Effective Health Care Program

Technical Brief
Number 23

Genetic Testing for Developmental Disabilities, Intellectual Disability, and Autism Spectrum Disorder

AHRQ
Agency for Healthcare Research and Quality
Advancing Excellence in Health Care • www.ahrq.gov

Technical Brief

Number 23

Genetic Testing for Developmental Disabilities, Intellectual Disability, and Autism Spectrum Disorder

Prepared for:
Agency for Healthcare Research and Quality
U.S. Department of Health and Human Services
540 Gaither Road
Rockville, MD 20850
www.ahrq.gov

Contract No. 290-2012-00011-I

Prepared by:
ECRI Institute–Penn Medicine Evidence-based Practice Center
Plymouth Meeting, PA

Investigators:
Fang Sun, M.D., Ph.D.
Jeff Oristaglio, Ph.D.
Susan E. Levy, M.D., M.P.H.
Hakon Hakonarson, M.D., Ph.D.
Nancy Sullivan, B.A.
Joann Fontanarosa, Ph.D.
Karen M. Schoelles, M.D., M.S., FACP

AHRQ Publication No. 15-EHC024-EF
June 2015

This report is based on research conducted by the ECRI–Penn Medicine AHRQ Evidence-based Practice Center (EPC) under contract to the Agency for Healthcare Research and Quality (AHRQ), Rockville, MD (290-2012-00011-I). The findings and conclusions in this document are those of the authors, who are responsible for its contents; the findings and conclusions do not necessarily represent the views of AHRQ. Therefore, no statement in this report should be construed as an official position of AHRQ or of the U.S. Department of Health and Human Services.

The information in this report is intended to help health care decisionmakers—patients and clinicians, health system leaders, and policymakers, among others—make well-informed decisions and thereby improve the quality of health care services. This report is not intended to be a substitute for the application of clinical judgment. Anyone who makes decisions concerning the provision of clinical care should consider this report in the same way as any medical reference and in conjunction with all other pertinent information, in the context of available resources and circumstances presented by individual patients.

AHRQ or U.S. Department of Health and Human Services endorsement of such derivative products that may be developed from this report, such as clinical practice guidelines, other quality enhancement tools, or reimbursement or coverage policies may not be stated or implied.

None of the investigators have any affiliation or financial involvement that conflicts with the material presented in this report.

Suggested citation: Sun F, Oristaglio J, Levy SE, Hakonarson H, Sullivan N, Fontanarosa J, Schoelles KM. Genetic Testing for Developmental Disabilities, Intellectual Disability, and Autism Spectrum Disorder. Technical Brief No. 23. (Prepared by the ECRI Institute–Penn Medicine Evidence-based Practice Center under Contract No. 290-2012-00011-I.) AHRQ Publication No.15-EHC024-EF. Rockville, MD: Agency for Healthcare Research and Quality; June 2015. www.effectivehealthcare.ahrq.gov/reports/final.cfm.

Preface

The Agency for Healthcare Research and Quality (AHRQ), through its Evidence-based Practice Centers (EPCs), sponsors the development of evidence reports and technology assessments to assist public- and private-sector organizations in their efforts to improve the quality of health care in the United States. The reports and assessments provide organizations with comprehensive, science-based information on common, costly medical conditions and new health care technologies and strategies. The EPCs systematically review the relevant scientific literature on topics assigned to them by AHRQ and conduct additional analyses when appropriate prior to developing their reports and assessments.

This EPC evidence report is a Technical Brief. A Technical Brief is a rapid report, typically on an emerging medical technology, strategy or intervention. It provides an overview of key issues related to the intervention—for example, current indications, relevant patient populations and subgroups of interest, outcomes measured, and contextual factors that may affect decisions regarding the intervention. Although Technical Briefs generally focus on interventions for which there are limited published data and too few completed protocol-driven studies to support definitive conclusions, the decision to request a Technical Brief is not solely based on the availability of clinical studies. The goals of the Technical Brief are to provide an early objective description of the state of the science, a potential framework for assessing the applications and implications of the intervention, a summary of ongoing research, and information on future research needs. In particular, through the Technical Brief, AHRQ hopes to gain insight on the appropriate conceptual framework and critical issues that will inform future research.

AHRQ expects that the EPC evidence reports and technology assessments will inform individual health plans, providers, and purchasers as well as the health care system as a whole by providing important information to help improve health care quality.

We welcome comments on this Technical Brief. They may be sent by mail to the Task Order Officer named below at: Agency for Healthcare Research and Quality, 540 Gaither Road, Rockville, MD 20850, or by email to epc@ahrq.hhs.gov.

Richard Kronick, Ph.D.
Director
Agency for Healthcare Research and Quality

David Meyers, M.D.
Acting Director
Center for Evidence and Practice Improvement
Agency for Healthcare Research and Quality

Stephanie Chang, M.D., M.P.H.
Director
Center for Evidence and Practice Improvement
Agency for Healthcare Research and Quality

Suchitra Iyer, Ph.D.
Task Order Officer
Center for Evidence and Practice Improvement
Agency for Healthcare Research and Quality

Acknowledgments

The authors thank Eileen Erinoff for her assistance in literature search, Janice Kaczmarek and Gina Giradi for their help in coordinating the project, Eloise DeHaan for copyediting, and Lydia Dharia and Kitty Donahue for document formatting and reference management. We also thank Dr. Craig A Umscheid, Senior Associate Director of ECRI–Penn Medicine AHRQ Evidence-based Practice Center (EPC) for recruiting clinical investigators. We would like to thank the Key Informants, Peer Reviewers, Task Order Officer, and Associate Editor, Dr. Margaret A. Piper, of the Kaiser Permanente Research Affiliates Evidence-based Practice Center, for their invaluable input. We would also like to thank the staff of the National Center for Biotechnology Information (NCBI) Genetic Testing Registry (GTR) for providing us with data on genetic tests.

Key Informants

In designing the study questions, the EPC consulted a panel of Key Informants who represent subject experts and end-users of research. Key Informant input can inform key issues related to the topic of the technical brief. Key Informants are not involved in the analysis of the evidence or the writing of the report. Therefore, in the end, study questions, design, methodological approaches and/or conclusions do not necessarily represent the views of individual Key Informants.

Key Informants must disclose any financial conflicts of interest greater than $10,000 and any other relevant business or professional conflicts of interest. Because of their role as end-users, individuals with potential conflicts may be retained. The TOO and the EPC work to balance, manage, or mitigate any conflicts of interest.

The list of Key Informants who participated in developing this report follows:

Michele Caggana, Sc.D.
Wadsworth Center
New York State Department of Health
Albany, NY

Ellen Roy Elias, M.D.
Children's Hospital Colorado
Aurora, CO

Margaret Daniele Fallin, Ph.D.
Johns Hopkins School of Public Health
Baltimore, MD

Ian Krantz, M.D.
Perelman School of Medicine
University of Pennsylvania
Philadelphia, PA

Amy Poole-Yaeger, M.D.
Centene Corp.
St. Louis, MO

Cheryl Reid, M.D.
Horizon Blue Cross Blue Shield of New Jersey
West Trenton, NJ

Stuart Shapira, M.D., Ph.D.
National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention
Atlanta, GA

Peter J. Smith, M.D., M.A.
Associate Professor of Pediatrics
University of Chicago
Chicago, IL

Marc Williams, M.D., FAAP, FACMG
Genomic Medicine Institute
Geisinger Health System
Danville, PA

Peer Reviewers

Prior to publication of the final evidence report, EPCs sought input from independent Peer Reviewers without financial conflicts of interest. However, the conclusions and synthesis of the scientific literature presented in this report does not necessarily represent the views of individual reviewers.

Peer Reviewers must disclose any financial conflicts of interest greater than $10,000 and any other relevant business or professional conflicts of interest. Because of their unique clinical or content expertise, individuals with potential non-financial conflicts may be retained. The TOO and the EPC work to balance, manage, or mitigate any potential non-financial conflicts of interest identified.

The list of Peer Reviewers follows:

Raphael Bernier, Ph.D.
Associate Professor, Department of Psychiatry & Behavioral Sciences
University of Washington
Seattle, WA

Linda A. Bradley, Ph.D., FACMG
Women and Infants Hospital Rhode Island/Alpert Medical School
Brown University
Providence, RI

Edwin H. Cook Jr., M.D.
Department of Psychiatry
University of Illinois at Chicago
Chicago, IL

Karen W. Gripp, M.D.
Division of Genetics, Department of Pediatrics
Nemours Children's Health System
Wilmington, DE

Genetic Testing for Developmental Disabilities, Intellectual Disability, and Autism Spectrum Disorder

Structured Abstract

Background. Genetics research in recent decades has discovered numerous genetic variants that help explain the etiology of developmental disabilities (DDs). Genetic tests (e.g., array comparative genomic hybridization, sequencing) are rapidly diffusing into clinical practice for diagnosing DDs or, more often, for determining their genetic etiology. An urgent need exists for a better understanding of these tests and their clinical utility.

Purpose. This Technical Brief collects and summarizes information on genetic tests clinically available in the United States to detect genetic markers that predispose to DDs. It also identifies, but does not systematically review, existing evidence addressing the tests' clinical utility. This Brief primarily focuses on patients with idiopathic or unexplained DDs, particularly intellectual disability, global developmental delay, and autism spectrum disorder. Several better-defined DD syndromes, including Angelman syndrome, fragile X syndrome, Prader-Willi syndrome, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, and Williams syndrome are also included. Patient-centered health outcomes (e.g., functional or symptomatic improvement) and intermediate outcomes (e.g., changes in clinical decisions or family reproductive decisions, the tests' diagnostic accuracy and analytic validity) are examined.

Methods. We sought input from nine Key Informants to identify important clinical, technology, and policy issues from different perspectives. We searched the National Center for Biotechnology Information's Genetic Testing Registry (GTR) to identify genetic tests. A structured search of studies published since 2000 was performed to identify available evidence that addresses genetic tests' clinical utility.

Findings. Our search of the GTR database identified 672 laboratory-developed tests offered by 63 providers in 29 States. We also identified one test cleared by the U.S. Food and Drug Administration. Common genetic testing methods used include array comparative genomic hybridization, microarray, DNA sequencing (the Sanger method or next-generation sequencing), and polymerase chain reaction. We did not identify any studies that directly assessed the impact of genetic testing on health outcomes. Most of the clinical studies identified for indirect assessment of clinical utility are case series reporting on a test's diagnostic yield.

Contents

Tables

Figures

Appendixes

Appendix H. Appendix Reference List

Background

Recent decades have witnessed numerous advances in genetics research highlighting the importance of genetic factors as an etiology for developmental disabilities (DDs). Given the rapid diffusion of advanced genetic tests for diagnosing DDs or determining their etiology, the Agency for Healthcare Research and Quality (AHRQ) commissioned the ECRI Institute–Penn Medicine Evidence-based Practice Center to prepare this Technical Brief to provide an overview of these tests. The brief summarizes information on tests that are clinically available in the United States. The brief also identifies existing evidence addressing the clinical utility of genetic tests for DDs. An evidence map is presented to outline evidence gaps on the subject and provide guidance for future research.

Developmental Disabilities

DDs are a group of conditions associated with functional impairment in physical, learning, language, or behavior areas.[1] According to this definition, the Centers for Disease Control and Prevention (CDC) categorizes a broad range of conditions as DDs, such as attention-deficit/hyperactivity disorder, autism spectrum disorder (ASD), cerebral palsy, hearing loss, learning disability, intellectual disability (ID), Tourette syndrome, vision impairment, and others.[1] The prevalence of DDs is estimated to be more than 15 percent in children 3–17 years of age.[2] These disorders, which often require lifelong individual and family support or treatment, have a profound impact on patients, families, and society. DDs can be caused or influenced by a variety of genetic and environmental factors, including gene mutations, mother's health behaviors (e.g., smoking and drinking) and untreated illness (e.g., phenylketonuria), complications during pregnancy or birth, and the exposure of the pregnant mother or child to infections or environmental toxins.[1] The causes of some developmental disabilities (e.g., Down syndrome, fragile X syndrome, fetal alcohol syndrome) are well understood. However, the underlying causes of many other DDs (e.g., ASD, ID) are often unclear and may vary substantially across individuals.

DDs can affect cognitive (including language and non-verbal problem solving), motor, and/or sensory functions. This Technical Brief focuses on genetic tests for evaluating DDs with primarily cognitive impairments, particularly idiopathic, nonsyndromic ID, ASD, and global developmental delay (GDD). Additionally, several DD syndromes were also included in this report based on key stakeholders' interest. These syndromes include Angelman syndrome, fragile X syndrome, Prader-Willi syndrome, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, and Williams syndrome. For patients with these syndromes, manifestations of GDD, ID, or ASD might be the main reason for families to seek evaluation when overt dysmorphic features have not been noted. DDs primarily diagnosed based on sensory or motor impairments (e.g., cerebral palsy, hearing loss, vision impairment) are beyond the scope of this brief. The findings of this Technical Brief should be interpreted as applying only to the DD conditions included in the report.

Intellectual Disability and Autism Spectrum Disorders

ID is a DD that may present in infancy or early childhood years. The American Association on Intellectual and Developmental Disabilities (AAIDD) defines ID as "a disability characterized by significant limitations both in intellectual functioning and in adaptive behavior as expressed in conceptual, social and practical adaptive skills."[3] ID affects 1 percent to 3 percent of the

population worldwide,[4,5] and about 0.7 percent of children aged 3–17 years in the United States.[2] Clinically, ID is diagnosed using standardized measures of developmental skills. These measures cover the domains of intelligence (IQ), adaptive behavior, and language function.[6,7] Because these standardized measures may be less reliable and valid for children younger than 5 years of age, establishing the clinical diagnosis of ID in early childhood can be challenging.[3]

Some possible causes that have been linked to ID include genetic aberrations, exposure to harmful substances (e.g., alcohol) or infection during pregnancy, complications during birth, acquired brain injury, and preterm birth.[8] Managing ID includes family support, family education and counseling, and special educational programs that may begin as early as infancy.[9] The goal of treatment is to support the child and family to enable the child to develop to his full potential.

ASD is a complex neurodevelopmental disorder, characterized by impairment in social interaction and communication and restricted, repetitive, and stereotyped patterns of behavior. Symptoms and profiles of children with ASD vary. In the period from 2006 to 2008, studies reported that autism affected 0.74 percent of children aged 3–17 years in the United States.[2] In 2010, the Autism and Developmental Disabilities Monitoring Network sites reported that the prevalence of ASD among children aged 8 years was 1.47 percent.[10] Both genetic and environment factors (e.g., maternal valproic acid use during pregnancy, congenital rubella) may play a role in ASD development,[11] and multiple genes have been found to influence ASD risk.[12]

ASD diagnosis is based on interviews with the child and family, a review of records and historical information, and examination of the child using standardized instruments (e.g., Autism Behavior Checklist, Autism Diagnostic Interview-Revised, Autism Diagnostic Observation Schedule) to demonstrate presence of core features of ASD. The results of clinical evaluation are compared with the diagnostic criteria included in the fifth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-5).[13] The diagnosis of ASD in infants and very young children is more difficult because developmental and behavioral assessment of these children may be more challenging. Some core features (e.g., socialization deficits, repetitive behavior, stereotyped movements) may emerge later as the child develops.

Key treatments for children with ASD include structured educational and behavioral interventions (e.g., applied behavioral analysis) to address core and associated symptoms and to promote development of social and communication skills. Behavioral interventions and medication may be used to address comorbid symptoms (e.g., anxiety, depression) or severe behavioral problems (e.g., aggression, self-injury).

Because establishing a diagnosis of ID or ASD may be challenging in infants and very young children, the term “global developmental delay” (GDD) is often used to categorize children who are younger than 5 years of age who have a significant delay in two or more developmental domains, including gross or fine motor, speech and language, cognitive, social and personal, and activities of daily living.[3,14] Significant delay is defined as performance two standard deviations or more below the mean on age-appropriate, standardized, normal-referenced testing.[15] Evaluating developmental delays requires accurate documentation by using norm-referenced and age-appropriate standardized measures of development administered by experienced developmental specialists.[3,15] Although GDD suggests a possible future diagnosis of ID and/or ASD, a child with GDD is not necessarily destined to have those conditions.

The prevalence of GDD is unknown, but may be similar to that of ID and ASD. GDD has a heterogeneous etiologic profile and is associated with age-specific deficits in adaptation and learning skills. Both genetic and environmental factors may be associated with GDD.[15,16]

Genetic Testing for Developmental Disabilities

Genetic abnormalities have been linked to many DDs. Studies suggest that up to 40 percent of DDs may be caused by some genetic aberration.[17,18] Conventional G-banded karyotyping has been used for decades to confirm the diagnosis of DDs (e.g., aneuploidies) that have a well-defined genetic etiology. More recently, new genetic methods (e.g., microarray-based comparative genomic hybridization [aCGH], whole genome or exome sequencing) have been developed and used to detect genetic abnormalities associated with DDs. These newer tests support the examination of genetic information at a higher resolution and may show genetic abnormalities not seen on G-banded karyotyping. In Appendix B, we provide a detailed technical overview to help illustrate how these genetic testing methods work and the main differences between them.

As previously discussed, clinical diagnosis of ID, ASD, or GDD is typically based on clinical manifestations and cognitive and developmental assessment using standardized measures. ID, ASD, and GDD are "functional diagnoses," which are phenotype-oriented descriptions of DDs. Each of these functional diagnoses includes multiple disorders of different etiologies (e.g., Angelman syndrome, fragile X syndrome, Prader-Willi syndrome, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, and Williams syndrome). When genetic tests are used to assess patients diagnosed with ID, ASD, or GDD, they are not used to confirm these functional diagnoses. Instead, these tests are used to establish an "etiologic diagnosis," that is, whether a patient who has an apparent functional diagnosis carries a specific genetic variant.

Etiologic diagnosis is a genotype-oriented description of genetic disorders and may be viewed as an early stage of defining a clinical disorder that has not yet been well understood or defined. A new etiologic diagnosis (i.e., the new genetic variant identified) can be further evaluated among individuals with the genotype in common to determine whether or not they share a common phenotype. If they share a common phenotype, a new genetic disorder may be defined and the genotype becomes part of the clinical definition of the disorder (e.g., fragile X syndrome).

Proposed benefits of establishing an etiologic diagnosis in patients with ID, ASD, or GDD include the following:[3,19-26]

- Clarifying a genetic cause and improving the psychosocial outcomes (e.g., improved knowledge and sense of empowerment) for patients and their families
- Providing prognosis or expected clinical course
- Evaluating recurrence risks and helping families in reproductive decisionmaking
- Refining treatment options
- Avoiding unnecessary and redundant diagnostic tests
- Identifying associated medical risks to prevent morbidity
- Providing condition-specific family support
- Facilitating acquisition of needed services and improving access to research treatment protocols

Because of these potential benefits, genetic tests are being used at an increasingly rapid rate. Medical genetics groups now recommend chromosomal microarray analysis (CMA) as a first-line genetic test to identify genetic mutations in children with multiple anomalies not specific to well-delineated syndromes, nonsyndromic DD/ID, and ASD.[3,19,21,27] Payers have seen a significant number of claims for genetic testing in children with suspected or proven DDs.[28]

However, little evidence from controlled studies exists to directly link genetic testing to health outcomes.[29] Published studies have reported superior diagnostic yields of newer genetic tests (e.g., aCGH) in identifying DD-related genetic abnormalities, and some have identified the impact of the tests on medical management (e.g., medical referrals, diagnostic imaging, further laboratory testing).[20-26] However, these findings are not sufficient for drawing a conclusion that use of the tests will lead to improved health outcomes (further discussion on this issue is provided in a later section, Establishing the Clinical Utility of Genetic Tests).

The impact of increased use of genetic tests, such as CMA, on health care costs is unclear. Advanced genetic tests are generally more expensive to perform than conventional G-banded karyotyping or other clinical tests.[30] Identification of genetic abnormalities on germline cells may also lead to genetic testing in patients' relatives, which further expands the pool of children for testing and magnifies the potential cost impact. Conversely, the potential increased diagnostic yield of advanced genetic tests may reduce the number of other clinical tests or services used to identify genetic causes of DDs. Besides the uncertain clinical utility and concerns about economic impact, ethical issues—such as how to deal with genetic abnormalities unrelated to DD that are detected in genome-wide CMA—also remain controversial.[31]

Availability of Genetic Tests for Developmental Disabilities in the United States

Genetic tests become clinically available in the United States via one of two pathways. A genetic test may reach the market as a commercially distributed test kit approved or cleared by the U.S. Food and Drug Administration (FDA) or as a laboratory-developed test (LDT).[32,33] Test kits cleared or approved by FDA include all reagents and instructions needed to complete the test procedure and interpret the results. These test kits can be used in multiple laboratories. LDTs are developed in laboratories using either FDA-regulated or self-developed analyte-specific reagents and are intended for performance solely in the test developer's laboratory.

The U.S. Centers for Medicare & Medicaid Services regulates laboratories that perform LDTs under the Clinical Laboratory Improvement Amendments of 1988 (CLIA).[32,33] Under CLIA regulations, facilities that perform tests on "materials derived from the human body for the purpose of providing information for the diagnosis, prevention, or treatment of any disease or impairment of, or the assessment of the health of, human beings" must obtain a certificate from the CLIA program. The requirements for CLIA certification are based on the complexity of the tests. LDTs compose the majority of the genetic tests that have become available to clinical practice.[33] Laboratories offering LDTs must be licensed as high-complexity clinical laboratories under CLIA regulations. A technology assessment report suggested that genetic tests for diagnosing DDs are primarily available as LDTs.[29]

Historically, FDA has exercised regulatory enforcement discretion over LDTs because they were relatively simple lab tests. As LDTs become more complex and proliferate in clinical use, the agency is taking steps to actively regulate LDTs. On October 3, 2014, FDA published two draft guidance documents regarding oversight of LDTs, titled "Framework for Regulatory Oversight of Laboratory Developed Tests (LDTs)" and "FDA Notification and Medical Device Reporting for Laboratory Developed Tests (LDTs)."[34,35] Under the proposed regulatory framework, LDTs will fall into one of the three categories: LDTs subject to full enforcement discretion; LDTs subject to partial enforcement discretion; and LDTs subject to full FDA regulation. Once the proposed FDA guidance documents are finalized, it will become clearer how genetic tests for DDs will be regulated.

Evaluating the Clinical Utility of Genetic Tests

The clinical utility of a medical test refers to how likely the test is to affect clinical decisions and ultimately improve patient outcomes. The ideal type of evidence for establishing clinical utility is from high-quality randomized controlled trials (RCTs) that compare health outcomes in patients who undergo the test to those in patients who do not. In reality, however, this type of RCT is rarely conducted.[32,33,36] To answer the ultimate clinical utility question—whether use of the test will improve health outcomes—an inference-based chain of evidence often needs to be established.[36,37] Establishing this chain of evidence involves assessing the analytic validity and clinical validity of the test of interest and establishing an indirect evidence link to clinical outcomes.

Analytic validity refers to how accurately and reliably the test measures the analyte of interest, such as a gene or chromosomal variant. Analytic validity is a function of many factors such as analytic accuracy, precision, analytic sensitivity and specificity, robustness, and reference range or normal values. The technical terms for analytic validity are defined in Appendix C of this report, Definition of Terms.

Clinical validity, also known as diagnostic accuracy, refers to how accurately the test detects or predicts the clinical condition of interest. Clinical validity is usually described in terms of clinical sensitivity, clinical specificity, positive and negative predictive values, likelihood ratios, diagnostic odds ratios, and the area under a receiver operator characteristic (ROC) curve. These technical terms related to clinical validity are also defined in Appendix C of this report. To address clinical validity, diagnostic cohort studies that use a gold-standard reference method are ideal.

When direct evidence does not exist or is insufficient to draw a reliable conclusion on whether testing improves health outcomes, addressing clinical validity and analytic validity may provide indirect evidence on the likelihood of a test affecting health outcomes. For example, if evidence shows that a test does not have adequate analytic validity, then the test is not likely to have adequate clinical validity. If a test does not have adequate clinical validity, it will have limited impact on treatment or family decisions. If a test does not change clinical or family decisions, it is unlikely to affect health outcomes.

Guiding Questions and Scope of Work

To meet this Technical Brief's objectives, we used a series of questions to guide our efforts in collecting appropriate information. These guiding questions fall into four categories:

1. Description of genetic tests for diagnosing or determining the etiology of DDs
    a. What genetic tests for diagnosing or determining the etiology of DDs are available for clinical practice in the United States?
    b. What genetic techniques or analysis methods (e.g., CMA, subtelomeric fluorescence in situ hybridization [FISH], whole genome or exome sequencing) are used in these tests? How do these types of techniques or methods work?
2. The context in which genetic tests are used for diagnosing or determining the etiology of DDs:
    a. What is the regulatory status (i.e., FDA clearance or approval status, CLIA certification of the test provider) of the tests?
    b. What kinds of credentials (i.e., training, certification) are required for interpreting test results?

c. Who are the providers ordering the tests and using their results?

3. State of the evidence on genetic tests for diagnosing or determining the etiology of DDs
    a. What are the DD conditions addressed in studies of these tests?
    b. What are the study designs used?
    c. What outcomes are reported?
        i. What data have been reported in the literature about the analytic validity of the tests?
        ii. What data have been reported in the literature about the clinical validity of the tests?
        iii. What data have been reported in the literature about the clinical utility of the tests?
        iv. What are the potential safety issues or harms related to the tests?
4. What are the important issues raised by genetic tests for diagnosing or determining the etiology of DDs?
    a. What are the proposed advantages and disadvantages of these tests compared with standard-of-care diagnostic methods?
    b. What recommendations do clinical practice guidelines include regarding the use of the tests?
    c. Given the current evidence status, what are the implications of the tests in terms of ethics, privacy, equity, cost, or economic efficiency?
    d. What are the current evidence gaps and potential areas of future research?
    e. What ongoing clinical trials are evaluating the clinical utility of the tests?
    f. What genetic tests or testing methods under research may become clinically available for diagnosing DDs in the near future?

The scope of work for this Technical Brief is described below by the population, interventions, comparators, and outcomes of interest. This scope reflects the ECRI Institute–Penn Medicine EPC team's current thinking and incorporates the input from AHRQ, the Technical Brief's topic nominators, the Key Informants, and Peer Reviewers.

Population: Children with DDs (e.g., ID and ASD) and their families (e.g., their siblings who may have the same disorder)

As previously discussed, this Technical Brief primarily focuses on patients with idiopathic or unexplained DDs, particularly IDs, ASD, or GDDs. These patients have clinical manifestations but may not have shown any distinct dysmorphic or syndromic features. Several better-defined DD syndromes are also included in the brief. These syndromes include Angelman syndrome, fragile X syndrome, Prader-Willi syndrome, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, and Williams syndrome. DDs that are typically diagnosed based upon motor or sensory impairments (e.g., cerebral palsy, hearing loss, vision impairment) or based on conventional G-banded karyotyping (e.g., aneuploidies) are beyond the scope of this report.

Interventions: Genetic tests for diagnosing DDs

This Technical Brief includes only tests that are available in the United States, either as FDA-cleared or FDA-approved test kits or as an LDT provided by a CLIA-certified laboratory. We primarily focus on CMA, including aCGH and single nucleotide polymorphism (SNP) assays, because use of these tests is widespread in clinical practice and because medical genetics

groups have recommended the tests for identifying genetic variants (e.g., copy number variations) in children with DDs.

Other types of genetic tests within the scope of work include polymerase chain reaction (PCR)-based tests (e.g., quantitative PCR); multiplex ligation-dependent probe amplification; Southern blot; whole genome or exome sequencing (e.g., using the Sanger method or next-generation sequencing); FISH, including subtelomeric FISH; and tests used for methylation analysis, deletion/duplication analysis, and uniparental disomy study. Conventional G-banded karyotyping is beyond the scope of work for this report.

Comparators: Comparators may vary across DDs and assessment purposes (e.g., clinical utility, clinical validity). For example, for assessing the clinical utility of a genetic test for determining the genetic etiology of ID, an appropriate comparator can be the standard-of-care diagnostic method without genetic testing. The standard-of-care method may include a physical examination by a clinician, a developmental assessment using standardized instruments (e.g., Wechsler Preschool and Primary Scale of Intelligence, Wechsler Intelligence Scales for Children, and Stanford-Binet Intelligence Scales), and other diagnostic tests (e.g., neuroimaging, metabolic studies).[28]

Outcomes: Patient-centered health outcomes and intermediate outcomes including changes in clinical decisions (e.g., refining treatment options, ordering other tests, referring patients to other specialists) or family reproductive decisions, diagnostic accuracy (e.g., sensitivity, specificity, positive and negative predict values), and parameters for measuring the analytic validity of a test.

Relevant outcomes may vary across different DDs. For example, for ASD, relevant outcomes may include reduction in autism symptom severity and improvement in language or adaptive behavior measured by a validated or standardized instrument (e.g., the Autism Diagnostic Observation Schedule, the MacArthur-Bates Communicative Development Inventories, Vineland Adaptive Behavior Scales). Outcomes relevant to ID include changes in cognition, remote memory, problem solving, understanding of relationships, social interaction, communication, self-care, and activities of daily living.

Methods

We describe below the methods for addressing the guiding questions previously defined.

Discussions with Key Informants

Within the Technical Brief process, Key Informants (KIs) serve as a resource to offer insight into the clinical context of the technology/intervention, how it works, how it is used or might be used, and which features may be important from a patient standpoint. The input of KIs were particularly important for this Technical Brief, because the area of genetic testing for developmental disabilities (DDs) is complex and published data for addressing some of the guiding questions are unavailable. The KIs helped identify relevant data sources and contributed to a better understanding of how advanced genetic tests work, their role in clinical practice, and potential advantages or harms. The KIs who worked with us for this project include clinicians who treat patients with DD, experts on genetic testing, patient advocates, medical directors from Medicaid programs, and individuals representing professional societies. Discussions with these KIs allowed us to identify important issues from different perspectives. Office of Management and Budget clearance was not required because we limited our standardized questions to no more than nine nongovernment-associated individuals.

After review and approval by the Agency for Healthcare Research and Quality (AHRQ) of the completed Disclosure of Interest forms for proposed KIs, we held interviews with nine selected KIs. The interviews were held with small groups of KIs based on availability and concordance of perspectives. Each interview was summarized in writing. The KIs' input was considered as we defined the project's scope of work and prepared the draft report. Information gained from KI interviews is identified as such in the report.

Gray Literature Search

A main objective of this Technical Brief is providing information on genetic tests clinically available for diagnosing or determining the etiology of DDs (refer to Guiding Questions 1 and 2). As discussed, the majority of these tests are available as LDTs. Identifying all LDTs within the scope of this Technical Brief has been a challenge and required a multi-faceted approach, including a comprehensive search of peer-reviewed and gray literature. Based on our experience in developing an Evidence-based Practice Center horizon scan report on molecular LDTs[33] in addition to the KIs' input, we used gray literature sources, particularly the National Center for Biotechnology Information (NCBI) Genetic Testing Registry (GTR) (www.ncbi.nlm.nih.gov/gtr/), as the primary source for identifying tests of interest.

NCBI is a division of the National Library of Medicine at the National Institutes of Health. The NCBI's GTR is a comprehensive information source for testing offered worldwide for disorders with a genetic basis.[38] Information is voluntarily submitted by test providers. Each test is assigned a stable identifier of the format GTR000000000. The GTR is designed to capture information on each test (e.g., its purpose, target populations, methods, what it measures, analytical validity, clinical validity, clinical utility, ordering information) and laboratory (e.g., location, contact information, certifications, licenses). However, the voluntarily submitted information is not equally complete for all data elements. For example, data on tests' analytical validity, clinical validity, or clinical utility are often missing. When these data were available, the sources of the data were rarely provided. In contrast, information on the laboratories that offer genetic tests are mostly complete.

According to its Web site (www.ncbi.nlm.nih.gov/gtr/), GTR requires test providers to review their data annually. Laboratory and test records that are one year beyond the last date they were edited will continue to display on the GTR site but are marked as past annual review. Laboratory and test records that are two years beyond the last date they were edited will be removed from GTR's public site.

We contacted GTR to request data on genetic tests. To fulfill our request, the GTR staff used the variables we provided and delivered the data in a Microsoft Excel file. We identified tests of interest using key terms for DD conditions (including their synonyms), GTR condition identifiers, and common genes known to be related to the conditions.

We also searched two other U.S.-focused online sources—McKesson Diagnostics Exchange and GeneTests.org—to complement and confirm the information collected from GTR. McKesson Diagnostics Exchange (https://app.mckessondex.com) is an online registry of molecular diagnostic tests. GeneTests.org (www.genetests.org) is a medical genetics information resource including a directory of international laboratories offering genetic testing. Both McKesson Diagnostics Exchange and GeneTests.org are proprietary but accessible by the public. Additional gray data sources we searched include GeneReviews® (www.ncbi.nlm.nih.gov/books/NBK1116/), the Association for Molecular Pathology Test Directory (www.amptestdirectory.org/index.cfm), NCBI's Online Mendelian Inheritance in Man® (OMIM) database (http://omim.org/), and EuroGentest (www.eurogentest.org).

We further searched other gray literature sources, such as government and specialty society Web sites, clinical trial databases, AHRQ's Healthcare Horizon Scanning System, trade publications, and meeting abstracts to identify data addressing these tests' analytic validity, clinical validity, and clinical utility (refer to Guiding Question 3). We also searched professional societies' Web sites to identify health technology assessment reports, clinical guidelines, and consensus statements, and new tests under development (to address Guiding Question 4).

Our search of the gray literature sources—except for the GTR—only identified a small amount of data on genetic tests. These data are less comprehensive and provide less detail than those identified from the GTR. The additional value of incorporating data from other sources to the data we had collected from the GTR was deemed limited. Methods that allow us to link data from different sources also are lacking. As a result, we decided to rely only on the GTR data for this Technical Brief.

Published Literature Search

We used a variety of databases to search the peer-reviewed literature, particularly for addressing Guiding Questions 3 and 4. We searched Medline and Embase using Embase.com, and PreMedline and the PubMed in-process subset through the National Library of Medicine Web site. We also searched PsycINFO (OVID) and the Cochrane library (including the Central Register of Controlled Trials, the Cochrane Database of Methodology Reviews, the Cochrane Database of Systematic Reviews, the Database of Abstracts of Reviews of Effects, the Health Technology Assessment Database, and the U.K. National Health Service Economic Evaluation Database). The National Guideline Clearinghouse (NGC) was searched for relevant clinical practice guidelines. The searches used a combination of controlled vocabulary terms and free-text words and were limited to English language studies published from January 2000 to January 2015. A detailed literature search strategy is provided in Appendix A.

Literature Review and Data Abstraction

Because of the broad scope of this Technical Brief (multiple DDs, multiple genetic tests, and various measures of test performance—analytic validity, clinical validity, and clinical utility), we screened and reviewed a large body of literature. Given the limited timeframe of this Technical Brief, a complete review of all full-text articles was not feasible. We therefore collected a portion of the data for this report from a review of abstracts. Given the nature of the data collected, this approach was sufficient for most studies. For example, the vast majority of clinical studies were case series that reported diagnostic yield for a given genetic target or the prevalence of a given genetic aberration among specific patient populations. In such instances, key information such as the study design (i.e., case series) and the reported outcome (e.g., diagnostic yield) were identified with confidence at the abstract level. In cases in which abstracts provided insufficient information, or there was reasonable uncertainty regarding the required data, we retrieved and reviewed full-text articles.

We collected data only from studies published in English that met the inclusion criteria specified in the Scope of Work section of this document. Data collection was guided by the criteria specified on data collection forms included in the Technical Brief Protocol submitted to AHRQ for review before we started the study. Data review and abstraction was performed with DistillerSR®, a Web-based systematic review software system (Evidence Partners, Inc., Ottawa, Ontario, Canada). Reviews of each abstract or full-text article and data extraction for each study was performed independently by two researchers. Any discrepancies between the two researchers regarding the selection or review of a given study were resolved through discussion or through arbitration led by a third researcher. Redundancies from multiple publications of the same data sets were identified and eliminated by reviewing author affiliations, study design, enrollment criteria, and enrollment dates. In such cases, the most recently published studies of these data were included in our report.

Findings

The findings of this Technical Brief are organized in the order of the Guiding Questions.

Genetic Tests for Developmental Disabilities (Guiding Questions 1 and 2)

Our search of the National Center for Biotechnology Information (NCBI) Genetic Testing Registry (GTR) database identified 672 tests (each assigned a unique GTR identifier) within the scope of this Technical Brief. These tests are offered by 63 providers located in 29 states (see Table 1). They are used for diagnosing, screening, or assessing the risk of developmental disabilities (DDs).They are all registered as "clinical tests" with "how to order" information. As we previously discussed, genetic tests are clinically available in the United States either as a FDA-cleared or -approved test or a laboratory-developed test (LDT). All the GTR tests identified are deemed to be LDTs, because the only FDA-cleared genetic test for developmental disorders (discussed later in this section) is not among these GTR tests.

The tables in Appendix D include detailed information about the GTR tests identified. The information is organized by the 11 DD categories, including intellectual disability (ID), autism spectrum disorder (ASD), global developmental delay (GDD), Angelman syndrome, fragile X syndrome, Prader-Willi syndrome, Rett syndrome, Rubinstein-Taybi syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, and Williams syndrome. Some tests are used for more than one DD category (e.g., used for both ID and fragile X syndrome or for both ASD and Rett syndrome). In these cases, the tests are counted in all categories that apply. These tables include the following information for each test:

- GTR test identification number
- Test provider information (name, location, lab test identification number, CLIA number)
- Target chromosomal regions or genes
- Genetic method used for analysis
- Specimen source
- Whether pre- or post-test counseling is required
- Whether the test is included in any proficiency testing program

All the data identified from the GTR were voluntarily reported by laboratories. NCBI established its protocol to guide the data submission process.[38] GTR provides the following note on its Web site (www.ncbi.nlm.nih.gov/gtr/):

> NIH [National Institutes of Health] does not independently verify information submitted to the GTR; it relies on submitters to provide information that is accurate and not misleading. NIH makes no endorsements of tests or laboratories listed in the GTR. GTR is not a substitute for medical advice. Patients and consumers with specific questions about a genetic test should contact a health care provider or a genetics professional.

We did not independently verify the data's accuracy either. Readers should use caution when they interpret data from GTR.

In addition, we identified some GTR tests listed under certain DD categories that analyze a gene without an established association with the DDs. For example, some tests listed under the velocardiofacial syndrome category analyze the FBN1 gene. This gene does not have an established association with velocardiofacial syndrome. FBN1 testing is typically used for diagnosing Marfan syndrome. Laboratories registered these tests under the velocardiofacial

syndrome category most likely for the purpose of differential diagnosis. For this Technical Brief, we excluded FBN1 tests from the velocardiofacial syndrome category. We also excluded tests of the same nature from other DD categories.

Table 1, Table 2, and Table 3 are a summary of the genetic tests we identified from the GTR database. As Table 1 indicates, we identified more tests for ID (or mental retardation) than for any other DD category. Not every State has a laboratory that offers genetic tests for DDs. The impact of the laboratories' geographic distribution on genetic tests' availability is unclear, because patients may receive testing services from laboratories in other States. Some laboratories offer multiple tests for the same DDs (e.g., ID, ASD). These tests differ in gene markers targeted and analysis methods used.

Proficiency testing (PT, a program in which externally validated samples are sent to a laboratory to test its accuracy in performing testing using its usual methodology) is performed for only a portion of the tests identified. The percentage of the tests for which proficiency testing is performed varies across the 11 DD categories, ranging from 22 percent for Rubinstein-Taybi syndrome to 74 percent for Prader-Willi syndrome. These PT programs include formal programs (e.g., those sponsored by the American College of Medical Genetics and Genomics and the College of American Pathologists) and intra-laboratory sample exchange programs.

As summarized in Table 2, the common analysis methods used in these tests include comparative genomic hybridization (CGH), microarray, single nucleotide polymorphism (SNP) detection, next-generation sequencing (NGS), bidirectional or unidirectional Sanger sequence analysis, multiplex ligation-dependent probe amplification (MLPA) or other polymerase chain reaction (PCR)-based methods, Southern blot, and fluorescent in situ hybridization (FISH). Some tests use more than one method (e.g., using both microarray and NGS; see tables in Appendix D). Sequencing (including both NGS and Sanger analysis) are the most frequently used methods among the tests identified. In these data tables, we use the same terminology that the GTR uses to label genetic methods. Some of these methods (e.g., microarray and CGH) potentially overlap.

Table 3 summarizes the genetic targets analyzed by the tests identified. GTR reports these target as a single gene (e.g., *PTEN*, *FMR1*, *UBE3A*, *MECP2*), a chromosome (e.g., chromosome 15), a chromosomal region (e.g., 15q11-q13), or the whole genome or exome (labelled as "human genome" in the GTR database). The most common targets in single-gene tests vary across DD categories (see Table 3). For the genes included in the multiple-gene tests, see tables in Appendix D.

In the tables in Appendix D, we reported information submitted to the GTR by test providers as to whether genetic counseling is required before or after genetic testing. We did not identify any information addressing credentials required for interpreting test results or data profiling providers who have ordered and used the tests.

Our search of the GRT database identified a limited amount of data on analytic validity or clinical validity for a portion of the 672 tests. However, references were rarely provided for determining where these data came from. We deemed these data to be unreliable and did not report them in this Technical Brief.

In addition to the tests we identified from GTR, we identified one U.S. Food and Drug Administration (FDA)-cleared commercial test kit that met the inclusion criteria for this Technical Brief. On January 17, 2014, the agency cleared Affymetrix CytoScan Dx Assay (Affymetrix, Inc., Santa Clara, CA) for marketing in the United States.[39] The test's FDA-cleared indication is below:[39]

> CytoScan Dx Assay is a qualitative assay intended for the postnatal detection of copy number variations (CNV) in genomic DNA obtained from peripheral whole blood in patients referred for chromosomal testing based on clinical presentation. CytoScan Dx Assay is intended for the detection of CNVs associated with developmental delay, intellectual disability, congenital anomalies, or dysmorphic features. Assay results are intended to be used in conjunction with other clinical and diagnostic findings, consistent with professional standards of practice, including confirmation by alternative methods, parental evaluation, clinical genetic evaluation, and counseling, as appropriate. Interpretation of assay results is intended to be performed only by healthcare professionals, board certified in clinical cytogenetics or molecular genetics. The assay is intended to be used on the GeneChip System 3000Dx and analyzed by Chromosome Analysis Suite Dx Software (ChAS Dx Software).
>
> This device is not intended to be used for standalone diagnostic purposes, preimplantation or prenatal testing or screening, population screening, or for the detection of, or screening for, acquired or somatic genetic aberrations.

According to the 510(k) clearance summary document filed with FDA, Affymetrix CytoScan Dx Assay uses chromosomal microarray technology and provides genome-wide coverage for detecting chromosomal imbalances.[39] The CytoScan Dx array contains 2.7 million markers that are representative of DNA sequences distributed throughout the genome. The majority of the markers (1.9 million) are nonpolymorphic markers. The assay reports the copy number state (loss, gain), copy number (i.e., 0, 1, 2, 3, or 4 or greater), and position or location of chromosomal segment copy-number changes across the queried genome.

Affymetrix had submitted data on CytoScan Dx Assay's performance to FDA for review. The submitted data addressed analytical performance (assay accuracy, precision/reproducibility, stability, assay controls, detection limit, analytical specificity, assay cut-off) and clinical sensitivity and specificity. These data were summarized in the FDA's 510(k) clearance summary document.[39]

Table 1. Summary of genetic tests: availability

Condition	Number of Tests Identified	Number of Laboratories Offering the Tests	Number of States Where Laboratories Are Located	States Where Laboratories Are Located	Number of Tests Participating in a PT Program
Angelman syndrome	82	39	24	California, Connecticut, Florida, Georgia, Illinois, Indiana, Massachusetts, Michigan, Minnesota, Mississippi, Missouri, Nebraska, New Jersey, New York, North Carolina, Ohio, Oklahoma, Oregon, Pennsylvania, South Carolina, Texas, Utah, Virginia, Wisconsin	45

Table 1. Summary of genetic tests: availability (continued)

Condition	Number of Tests Identified	Number of Laboratories Offering the Tests	Number of States Where Laboratories Are Located	States Where Laboratories Are Located	Number of Tests Participating in a PT Program
Autism spectrum disorders	93	23	16	California, Colorado, Connecticut, Florida, Georgia, Maryland, Massachusetts, Michigan, Nebraska, New York, Ohio, Pennsylvania, Texas, Utah, Virginia, Wisconsin	31
Fragile X syndrome	56	34	20	California, Colorado, Florida, Georgia, Iowa, Massachusetts, Michigan, Mississippi, Missouri, Montana, Nebraska, New York, Ohio, Oklahoma, Oregon, Pennsylvania, South Carolina, Texas, Utah, Wisconsin	30
Global developmental delay	27	18	13	California, Connecticut, Florida, Georgia, Illinois, Maryland, Massachusetts, Michigan, New York, Ohio, Texas, Utah, Virginia	17
Intellectual disability/ mental retardation	333	23	16	California, Colorado, Connecticut, Georgia, Illinois, Iowa, Massachusetts, Michigan, Nebraska, Ohio, Oklahoma, Pennsylvania, South Carolina, Texas, Utah, Wisconsin	114
Prader-Willi syndrome	50	39	24	California, Florida, Georgia, Illinois, Indiana, Iowa, Massachusetts, Michigan, Mississippi, Missouri, Montana, Nebraska, New Jersey, New York, North Carolina, Ohio, Oklahoma, Oregon, Pennsylvania, South Carolina, Texas, Utah, Virginia, Wisconsin	37

Table 1. Summary of genetic tests: availability (continued)

Condition	Number of Tests Identified	Number of Laboratories Offering the Tests	Number of States Where Laboratories Are Located	States Where Laboratories Are Located	Number of Tests Participating in a PT Program
Rett syndrome	100	24	18	California, Connecticut, Delaware, Florida, Georgia, Illinois, Massachusetts, Michigan, Mississippi, Missouri, Nebraska, Oklahoma, Oregon, Pennsylvania, South Carolina, Texas, Utah, Wisconsin	52
Rubinstein-Taybi syndrome	23	8	8	California, Colorado, Georgia, Illinois, Massachusetts, Nebraska, Ohio, Texas	5
Smith-Magenis syndrome	29	19	12	California, Connecticut, Georgia, Indiana, Massachusetts, Michigan, Nebraska, Ohio, Texas, Utah, Virginia, Wisconsin	14
Velocardiofacial syndrome	44	28	17	California, Connecticut, Florida, Georgia, Illinois, Indiana, Maryland, Massachusetts, Michigan, Montana, Nebraska, New York, Ohio, Texas, Utah, Virginia, Wisconsin	23
Williams syndrome	22	16	11	California, Georgia, Indiana, Massachusetts, Michigan, Nebraska, New York, Ohio, Utah, Virginia, Wisconsin	15

Source: National Center for Biotechnology Information's Genetic Testing Registry
PT=proficiency testing

Table 2. Summary of genetic tests: commonly used methods

Condition	CGH	Microarray	SNP Detection	NGS/MPS	Sanger Sequence Analysis	MLPA	PCR	FISH	Southern Blot	Methylation Analysis	Trinucleotide Repeats Analysis
Angelman syndrome	6	3	1	24	27	9	20	8	2	14	2
Autism spectrum disorders	11	16	7	29	30	9	6	1	4	0	4
Fragile X syndrome	3	3	0	7	5	0	42	0	34	5	33
Global developmental delay	1	13	7	7	2	0	1	0	0	0	0
Intellectual disability/ mental retardation	83	35	2	122	141	22	4	0	2	0	2
Prader-Willi syndrome	0	2	1	5	1	6	28	10	4	24	3
Rett syndrome	7	3	0	37	43	28	5	1	2	1	2
Rubinstein-Taybi syndrome	2	3	0	6	10	5	0	0	0	0	0
Smith-Magenis syndrome	2	3	0	12	5	0	0	6	0	0	0
Velocardiofacial syndrome	4	3	0	20	3	0	1	8	0	0	0
Williams syndrome	0	1	0	6	0	0	0	8	0	0	0

Source: National Center for Biotechnology Information's Genetic Testing Registry

Note: Some tests use more than one method.

CGH=comparative genomic hybridization; FISH=fluorescence in situ hybridization; MLPA=multiplex ligation-dependent probe amplification; NGS/MPS=next-generation sequencing/massively parallel sequencing; PCR=polymerase chain reaction; SNP=single nucleotide polymorphism

Table 3. Summary of genetic tests: genetic targets

Condition	Numbers of Single-Gene Tests	Common Target of Single-Gene Tests (Number of Tests)	Numbers of Tests Analyzing Multiple Genes	Numbers of Tests Analyzing a Chromosomal Region or the Whole Genome or Exome
Angelman syndrome	32	*UBE3A* (26), *MECP2* (6)	30	19
Autism spectrum disorders	43	*PTEN* (13), *MECP2* (7), *FMR1* (3)	32	15
Fragile X syndrome	46	*FMR1* (46)	8	2
Global developmental delay	4	*CTNND2* (1), *FGFR3* (1), *FMR1* (1), *GFER* (1)	8	15
Intellectual disability/ mental retardation	215	*MEF2C* (27), *ARX* (12), *CASK* (8), *HSD17B10* (8), *FKRP* (6), *OPHN1* (6)	116	2
Prader-Willi syndrome	24	*SNRPN* (22), *PWARSN* (1), *MAGEL2* (1)	5	21
Rett syndrome	59	*MEF2C* (41), *FOXG1* (18),	39	1
Rubinstein-Taybi syndrome	17	*CREBBP* (12), *EP300* (5)	6	1
Smith-Magenis syndrome	7	*RAI1* (7)	12	10
Velocardiofacial syndrome	5	TBX1 (3), HIRA (2)	20	18
Williams syndrome	1	*ELN* (1)	6	15

Source: National Center for Biotechnology Information's Genetic Testing Registry

Evidence for Addressing Clinical Utility (Guiding Question 3)

Figure 1 summarizes the literature review workflow identifying current evidence for addressing clinical utility. Our search of peer-reviewed journals and gray literature (including manual search of journal articles' reference lists) identified 2,242 records potentially relevant to the topic of this Technical Brief. We excluded 1,808 articles based on abstract review. Most of the articles were excluded because they were not about a clinical disorder or a genetic test of interest. We were able to extract data from 307 articles at the abstract level. We retrieved 127 full-length articles because their abstracts did not provide sufficient information for data extraction or for judging whether the articles were within the scope of work. We excluded 8 articles at the full-length article level. Then, we extracted data from 119 remaining full-length articles.

Ultimately, we extracted data from 426 studies at either the abstract or full-length article level. These data include the studies' design, reported outcomes, sample size, the performance aspects it addressed (e.g., analytic validity, clinical validity, clinical utility), targeted DDs (e.g., ASD, ID, fragile X syndrome), and testing methods used (e.g., PCR, sequencing). These data were exported from the Distiller system into an Excel file for analysis.

Table 4 to Table 9 together provide a map of evidence that directly or indirectly addresses the clinical utility of genetic tests for DDs. for this Technical Brief, we first searched for studies that directly addressed the clinical utility issues. The studies are summarized in Table 4. Our search did not identify any randomized controlled trials (RCTs) or non-RCT studies that directly

evaluated the impact of genetic testing on health outcomes. Our search identified seven studies that evaluated the impact of testing on clinical management (e.g., medical referrals, decisions on diagnostic imaging or other laboratory testing, improvement in acquiring services) or family decisions (e.g., reproductive decisions).[22,23,40-44] We also identified three studies that evaluated the value of genetic testing perceived by families affected by DDs.[25,44,45] These studies do not provide firm evidence regarding the influence of genetic testing on health outcomes, but they help estimate the possibility of an effect. Most identified studies evaluated chromosomal microarray analysis (CMA). Table 5 is a summary of the clinical utility studies organized by DD conditions. Many of these studies addressed genetic testing for multiple conditions. Developmental delay and ID were the most studied conditions.

Given that most of the genetic tests relevant to this report are intended to establish an etiologic diagnosis and rarely used in isolation to confirm a clinical diagnosis (e.g., ASD, ID, Angelman syndrome, velocardiofacial syndrome), we did not expect to identify many studies that reported outcomes addressing clinical validity, such as clinical sensitivity or specificity, clinical predictive values, and diagnostic odds ratios. In fact, we only found one study that addressed the clinical validity of a genetic test. Jiao and colleagues (2011) conducted a case-control study among 18 patients with ASD to evaluate the accuracy (sensitivity and specificity) of a diagnostic model based on SNPs and magnetic resonance imaging in predicting ASD subtypes.[46]

Our search identified 20 studies that addressed analytic validity issues (Table 6). These studies were intended to validate the analytic characteristics of one or more new tests for detecting genetic variants associated with DDs. Reported performance measures include analytic sensitivity or specificity, positive and negative predictive values, receiver operator characteristic (ROC) curves and area under the curve (AUC), reference range, repeatability, precision, concordance, and signal-to-noise ratio. The majority of the studies in Table 6 evaluated polymerase chain reaction (PCR) or chromosomal microarray analysis (CMA). They are either case-control studies or case series. Table 7 is a summary of the analytic validity studies organized by DD conditions. Many of these studies addressed genetic testing for multiple conditions. Fragile X syndrome was the most studied condition.

For this Technical Brief, we identified 129 case series that reported on the diagnostic yield of a genetic test. Diagnostic yield is calculated as the number of patients who had a "causal," "pathogenic," or "clinically significant" genetic aberration detected by the test, divided by the total number of patients tested. Although diagnostic yield indicates the percentage of patients being tested who ultimately reach a diagnosis, it does not reveal whether the diagnoses reached are correct or whether the targeted genetic aberration is truly causal, pathogenic, or clinically significant. Although improved diagnostic yield is often used by researchers as evidence to support the use of genetic tests, this improvement does not necessarily lead to improved health outcomes.

The diagnostic yield studies we identified are summarized in Table 8. These studies include those comparing diagnostic yields of two or more testing methods and those validating a new testing method. CMA was the most prevalent genetic test method (in 66 studies), followed by PCR (in 34 studies). Most of the diagnostic yield studies addressed multiple DD conditions. As Table 9 demonstrates, ID was the most studied DD (in 85 studies), followed by developmental delay (in 50 studies) and ASD (in 35 studies).

Additionally, we identified more than 200 studies (not including single-patient or single-family case reports) that investigated any association between a genetic marker (genotype) and a

DD disorder or its physical and mental characteristics (phenotype). These genotype-phenotype association studies are exploratory in nature. They were not intended to validate a genetic test. Instead, they used genetic tests as research tools. These studies generate hypotheses and provide valuable input for developing future genotype-phenotype associations. However, it is still premature to consider the findings of these studies in building the evidence chain for addressing genetic tests' clinical utility. Therefore, we do not report these studies in this Technical Brief.

Figure 1. Literature review workflow

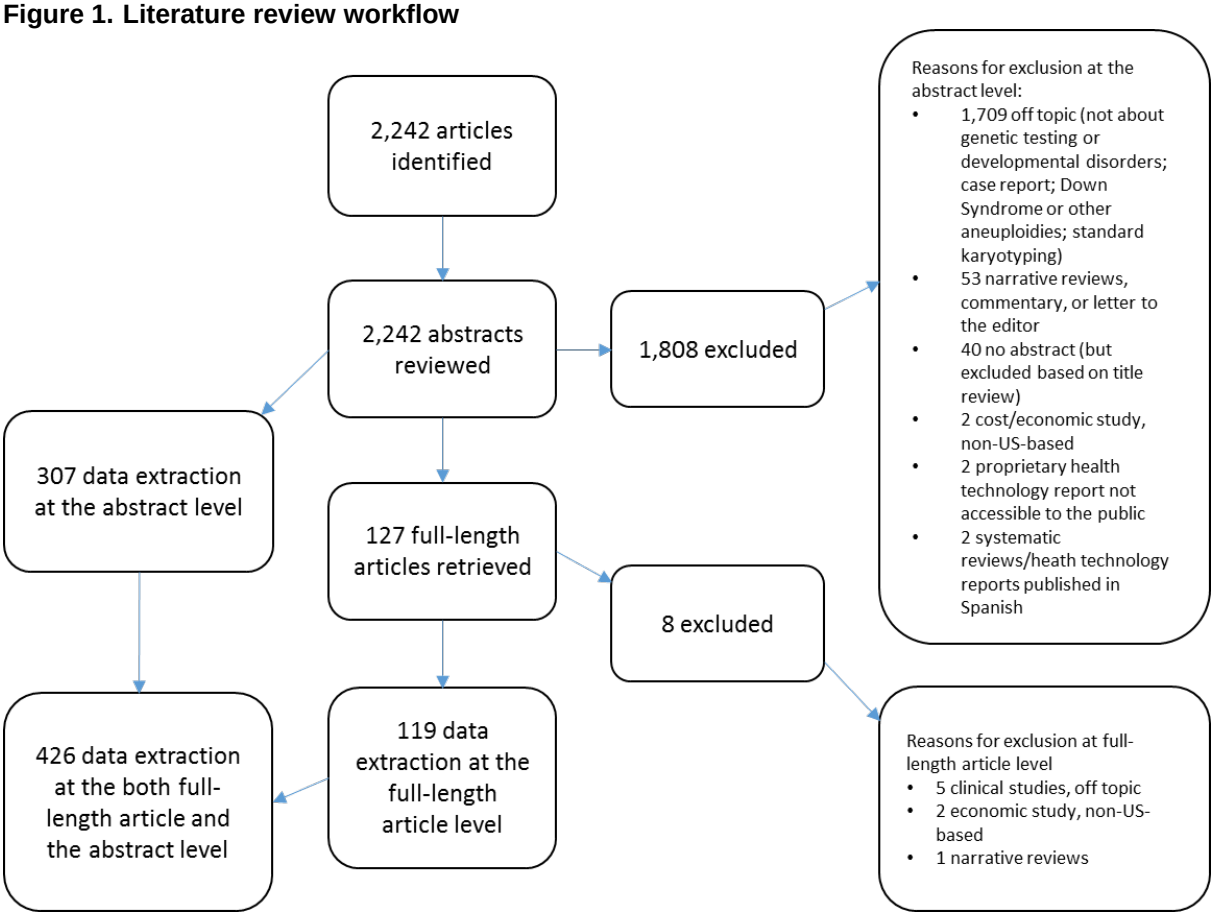

Table 4. Evidence map: clinical utility studies

Reference	DD Disorder	Test Studied	Study Design	Reported Outcomes	Sample Size
Srivastava et al. 2014[47]	ASD, DD, ID	Whole exome sequencing	Case series	Changes in clinical management	78
Amiet 2014[45]	ASD	CMA and FISH	Survey	Interest in the use of a genetic screening test for ASD	631
Ellison 2012[40]	DD, ID, Angelman syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, Williams syndrome	CMA	Case series	Changes in clinical management	122 (a subset of 46,298 cases)
Iglesias 2014[41]	DD, ID, ASD	Whole-exome sequencing	Case series	Changes in clinical or family decisions	115
Costain 2012[44]	Velocardiofacial syndrome	Molecular diagnosis	Survey	Impact on disease understanding and certainty, on advocacy, and on opportunities to optimize medical, social and educational needs	118
Mroch 2012[22]	DD, Rett syndrome	CMA	Case series	Changes in clinical management	3
Coulter 2011[23]	DD, ID, ASD	CMA	Case series	Changes in medical care by precipitating medical referrals, diagnostic imaging, or specific laboratory testing	1,792
Bruno 2009[42]	ID	CMA	Case series	Changes in clinical or family decisions	117
Makela 2009[43]	ID	CMA	Survey	Impact on experiences acquiring services, use of support groups, the family's reproductive decisions Interest regarding the importance of an etiological diagnosis	20
Saam 2008[25]	DD, Prader-Willi syndrome, muscular dystrophy	CMA	Survey	The willingness to pay for diagnostic testing to find a genetic cause of DD from families of children with DD	48

ASD=autism spectrum disorder; CMA=chromosomal microarray analysis; DD=developmental delay; FISH=fluorescence in situ hybridization; ID=intellectual disability

Table 5. Clinical utility studies by developmental disorders

DD Disorder	Number of Studies	Reference
Angelman syndrome	1	Ellison 2012[40]
Autism spectrum disorder	4	Amiet 2014[45], Srivastava et al. 2014[47], Iglesias 2014[41], Coulter 2011[23]
Developmental delay	6	Srivastava et al. 2014[47], Iglesias 2014[41], Ellison 2012[40], Mroch 2012[22], Coulter 2011[23], Saam 2008[25]
Intellectual disability	6	Iglesias 2014[41], Srivastava et al. 2014[47], Ellison 2012[40], Coulter 2011[23], Bruno 2009[42], Makela 2009[43]
Prader-Willi syndrome	1	Saam 2008[25]
Rett syndrome	1	Mroch 2012[22]
Smith-Magenis syndrome	1	Ellison 2012[40]
Velocardiofacial syndrome	1	Ellison 2012[40] Costain 2012[44]
Williams syndrome	1	Ellison 2012[40]

Table 6. Evidence map: analytic validity studies

Reference	DD Disorder	Analysis Methods Used in the Test Studied	Study Design	Reported Outcomes	Sample Size
Kalman 2014[48]	Rett syndrome	CMA, Sanger sequencing, PCR	Case series	Repeatability/precision	35
Inaba 2014[49]	Fragile X syndrome	PCR, methylation analysis	Case-control study	Sensitivity/specificity, Positive/negative predictive value	685
Hayes 2013[50]	Developmental delay	CMA, next-generation sequencing, FISH	Cohort study	Sensitivity/specificity	39
Stofanko 2013[51]	ID, Smith-Magenis syndrome	Quantitative fluorescent PCR	Case-control study	Sensitivity/specificity, positive/negative predictive value	428
Stofanko 2013[52]	ID, ASD, Angelman syndrome, Rett syndrome, velocardiofacial syndrome, Williams syndrome	Quantitative fluorescent PCR	Case-control study	Sensitivity/specificity, Positive/negative predictive value	29
Koshimizu 2013[53]	ASD	Next-generation sequencing	Case-control study, case series	Sensitivity/specificity	38
Lafauci 2013[54]	Fragile X syndrome	Luminex immunoassay	Case-control study	Sensitivity/specificity, ROC (AUC), repeatability/precision, reference range	215
Curtis-Cioffi 2012[55]	Fragile X syndrome	PCR-based screening, Southern blot	Cohort study	Sensitivity/specificity	78
Juusola 2012[56]	Fragile X syndrome	PCR-based detection	Case-control study	Sensitivity/specificity, repeatability/precision	76
Lessard 2012[57]	Fragile X syndrome	Western blot	Case-control study	Sensitivity/specificity, ROC (AUC), repeatability/precision	150
Bernardini 2010[58]	Developmental delay, ID, congenital abnormalities	CMA	Case-control study, case series	Sensitivity/specificity	70
Chen 2010[59]	Fragile X syndrome	Single-tube CGG repeat primed FMR1 PCR	Case series	Sensitivity	227
Filipovic-Sadic 2010[60]	Fragile X syndrome	FMR1 gene-specific PCR, Southern blot	Case-control study	Sensitivity/specificity, concordance	146
Coffee 2009[61]	Fragile X syndrome	Quantitative methylation-sensitive PCR, Southern blot	Case-control study	Sensitivity/specificity, positive/negative predictive value	36,124

Table 6. Evidence map: analytic validity studies (continued)

Reference	DD Disorder	Analysis Methods Used in the Test Studied	Study Design	Reported Outcomes	Sample Size
Hu 2009[62]	Fragile X syndrome	Next-generation sequencing, PCR	Case series	Sensitivity	24
Wang 2009[63]	Angelman syndrome, Prader-Willi syndrome	PCR and quantitative melting curve analysis	Case-control study	Ability to discriminate between deletional and nondeletional Prader-Willi and Angelman syndromes	143
Truong 2008[64]	Smith-Magenis syndrome	Real-time, quantitative PCR	Case-control study	Repeatability/precision, reported range	64
Ballif 2007[65]	Developmental delay, congenital abnormalities	CMA	Case series	Repeatability/precision	6,946
Shen 2007[66]	Developmental Delay, ID, ASD, Angelman syndrome, Smith-Magenis syndrome, velocardiofacial syndrome, Williams syndrome	CMA	Case-control study	Sensitivity/specificity, repeatability/precision, concordance, signal-to-noise ratio	316
Altug-Teber 2005[67]	Angelman syndrome, Prader-Willi syndrome	CMA	Case series	Sensitivity	6

ASD=autism spectrum disorder; CMA=chromosomal microarray analysis; FISH=fluorescence in situ hybridization; ID=intellectual disability; PCR=polymerase chain reaction; ROC (AUC)=receiver operator characteristic (area under the curve)

Table 7. Analytic validity studies by developmental disorders

DD Disorder	Number of Studies	Reference
Angelman syndrome	4	Stofanko 2013[52], Wang 2009[63], Shen 2007[66], Altug-Teber 2005[67]
Autism spectrum disorder	3	Koshimizu 2013[53], Stofanko 2013[52], Shen 2007[66]
Developmental delay	4	Hayes 2013[50], Bernardini , 2010[58], Ballif 2007[65], Shen 2007[66]
Fragile X syndrome	9	Inaba 2014[49], Lafauci 2013[54], Curtis-Cioffi 2012[55], Juusola 2012[56], Lessard 2012[57], Chen 2010[59], Filipovic-Sadic 2010[60], Coffee 2009[61], Hu 2009[62]
Intellectual Disability	4	Stofanko 2013[51], Bernardini 2010[58], Shen 2007[66], Stofanko 2013[52]
Prader-Willi syndrome	2	Wang 2009[63], Altug-Teber 2005[67]
Rett syndrome	2	Kalman 2014[48], Stofanko 2013[52]

Table 7. Analytic validity studies by developmental disorders (continued)

DD Disorder	Number of Studies	Reference
Smith-Magenis syndrome	3	Stofanko 2013[51], Truong 2008[64], Shen 2007[66]
Velocardiofacial syndrome	2	Stofanko 2013[52], Shen 200766
Williams syndrome	2	Stofanko 2013[52], Shen 2007[66]

Table 8. Case series reporting diagnostic yield

Reference	DD Disorder	Test Studied	Sample Size
Lee et al. 2014[68]	DD	Whole exome sequencing	138
Yang et al. 2014[69]	ASD, DD, ID	Whole exome sequencing	2,000
Pereira et al. 2014[70]	ID	CMA	15
Tao et al. 2014[71]	ASD, DD, ID	CMA	327
Soden et al 2014[72]	ASD, DD, ID and other neurodevelopmental disorders	Whole genome or exome sequencing	119
Srivastava et al. 2014[47]	ASD, DD, ID	Whole exome sequencing	78
Bartnik et al. 2014[73]	DD, ID, dysmorphic features; congenital anomalies	CMA	256
Boggula et al. 2014[74]	DD, ID, PW, SMS, VS, WS	FISH, MLPA	203
Byeon et al. 2014[75]	DD, ID	CMA, FISH, standard karyotyping	87
Chaudhary et al. 2014[76]	ID, FX	PCR (amplifying methylated CpG dinucleotide region or the unmethylated CGG repeats), Southern blot	63
Cheon et al. 2014[77]	ID, Kabuki syndrome	Whole exome and direct sequencing	12
Chong et al. 2014[78]	DD, ID, ASD	CMA	105
Coutton et al. 2014[79]	ID	CMA	66
Dubourg et al. 2014[80]	SMS	PCR, sequencing	9
Fatima et al. 2014[81]	ID, FX	PCR (using primers corresponding to modified methylated and unmethylated DNA), Southern blot	583
Gilissen et al. 2014[82]	ID	Whole-genome sequencing	50
Helsmoortel et al. 2014[83]	ID	CMA, whole-exome sequencing	10
Huguet et al. 2014[84]	ASD	Direct sequencing	1,578
Iglesias et al. 2014[41]	DD, ID, ASD	Whole exome sequencing	115
Kalman et al. 2014[48]	Rett	CMA, Sanger sequencing, semiquantitative PCR	35
Lee et al. 2014[85]	RTS	Sequencing, MLPA	16
Medina et al. 2014[86]	ID	MLPA	119
Nicholl et al. 2014[87]	DD, ID, ASD	CMA	1,700
Palmer et al. 2014[88]	ID	CMA	67
Pereira et al. 2014[70]	DD, ID	CMA	305
Stobbe et al. 2014[89]	ASD	CMA	36
Tucker et al. 2014[90]	ID	CMA	185
Tuysuz et al. 2014[91]	PW, hypotonia	FISH, methylation analysis; karyotyping	65
Utine et al. 2014[92]	ID	CMA	200
Uwineza et al. 2014[93]	DD, ID	CMA	50
Vergult et al. 2014[94]	ID, congenital malformations	CMA, FISH, mate-pair next-generation sequencing	50
Willemsen and Kleefstra 2014[95]	ID	Sanger sequencing	253
Wiszniewska et al. 2014[96]	DD, ID	CMA	3,240
Ahn et al. 2013[97]	AS, PW, SMS, VS	CMA	13,412
Battaglia et al. 2013[98]	DD, ID, ASD	CMA	349

Table 8. Case series reporting diagnostic yield (continued)

Reference	DD Disorder	Test Studied	Sample Size
Behjati et al. 2013[99]	ID	MLPA	100
Behjati et al. 2013[100]	ID	CMA, MLPA	32
Del Carmen et al. 2013[101]	VS	FISH	268
Doherty et al. 2013[102]	DD, FX	CMA, PCR, Southern blot	2,046
Esposito et al. 2013[103]	DD, ID, ASD, FX, facial dysmorphism	PCR, Southern blot	2,750
Essop and Krause 2013[104]	ID, FX	PCR, Southern blot	2,690
Fan et al. 2013[105]	DD, ASD	CMA	607
Halder et al. 2013[106]	AS, PW, VS	FISH	301
Hayes et al. 2013[50]	DD, learning difficulties	CMA, FISH, next-generation sequencing	39
Jain et al. 2013[107]	ID	CMA, karyotyping, molecular studies for FX	101
Jorge et al. 2013[108]	ID, FX	Multiplex PCR	100
Kashevarova et al. 2013[109]	DD, ID	CMA	71
Koshimizu et al. 2013[53]	ASD	Next-generation sequencing	38
Lee et al. 2013[110]	DD, ID	CMA	190
Marano et al. 2013[111]	DD, ID, ASD, seizures; dysmorphic features; hypotonia; failure to thrive	CMA	200
Mundhofir et al. 2013[112]	ID	MLPA	436
Nicholl et al. 2013[113]	DD, ID, ASD, congenital anomalies; epilepsy	CMA	247
Pohovski et al. 2013[114]	DD, ID	MLPA	150
Pratt et al. 2013[115]	Rett	PCR, Sanger sequencing, MLPA	12
Qiao et al. 2013[116]	DD, ID, ASD	CMA, quantitative multiplex PCR	82
Rodriguez-Revenga et al. 2013[117]	DD, ID	CMA, FISH, MLPA, quantitative PCR	200
Saad et al. 2013[118]	WS	FISH	17
Shoukier et al. 2013[119]	DD, ID, congenital anomalies	CMA	342
Sorte et al. 2013[120]	ASD	CMA	50
Tos et al. 2013[121]	ID	FISH	67
Vallespin et al. 2013[122]	DD, ID, ASD	Custom whole-genome oligonucleotide-based array (called KaryoArrayv3.0; Agilent-based 8 x 60 K)	780
Vallespin et al. 2013[123]	ID, ASD, multiple congenital anomalies	FISH, custom whole-genome oligonucleotide-based array; MLPA; karyotype	120
Vorsanova et al. 2013[124]	ID, ASD	CMA	100
Winarni et al. 2013[125]	ASD, FX	PCR, Southern blot, cytogenetic analysis	65
Zarate et al. 2013[126]	DD, ID, ASD, FX	CMA, FISH, quantitative PCR	59
Aradhya et al. 2012[127]	Mendelian disorders overall	CMA, DNA sequencing	3,018

Table 8. Case series reporting diagnostic yield (continued)

Reference	DD Disorder	Test Studied	Sample Size
Dos Santos and Freire-Maia 2012[128]	DD, ID	FISH	15
Ellison et al. 2012[40]	DD, ID, AS, PW, SMS, VS, WS	CMA	46,298
Hochstenbach et al. 2012[129]	ID	Multiplex enrichment and next-generation sequencing of the entire coding sequence of all genes	20
Iourov et al. 2012[130]	DD, ID, ASD	CMA	54
McGrew et al. 2012[131]	ASD, FX	CMA, fragile X DNA testing	259
Rafati et al. 2012[132]	ID	MLPA	328
Rafati et al. 2012[133]	ID	FISH, MLPA	322
Splendore et al. 2012[134]	Rett	Direct sequencing	139
Tos et al. 2012[135]	ID, multiple congenital anomalies	FISH, standard chromosomal analysis	24
Tzetis et al. 2012[136]	DD, ID, ASD, deafness, seizures, multiple congenital anomalies	CMA, FISH, "conventional karyotype"	334
Utine et al. 2012[137]	ID	MLPA	100
Bremer et al. 2011[138]	ASD	CMA	223
Bruno et al. 2011[139]	DD, ID, ASD, congenital anomalies	CMA	5,000
Coulter et al. 2011[23]	DD, ID, ASD, congenital anomalies	CMA	1,792
Hannibal et al. 2011[140]	ID	Sequencing	110
Hayashi et al. 2011[141]	ID, multiple congenital anomalies	CMA, conventional cytogenetics	536
Rana et al. 2011[142]	ID	MLPA technique for subtelomeric anomalies	35
Roesser 2011[143]	ASD	Karyotype, DNA for FX	507
Rooms et al. 2011[144]	ID, FX, "negative for FX"	Array-based MLPA	413
Shawky et al. 2011[145]	ID	FISH, routine conventional karyotyping, high resolution banding	30
Wincent et al. 2011[146]	DD	CMA	160
Bernardini et al. 2010[58]	DD, ID, congenital anomalies	CMA	70
Dave et al. 2010[147]	ID, FX	PCR, Southern Blot	720
Ezugha et al. 2010[148]	DD, ID, ASD, learning disability, hypotonia	CMA	82
Filipovic-Sadic et al. 2010[60]	FX	FMR1 gene-specific PCR, Southern blot	146
Gervasini et al. 2010[149]	DD, RTS	CMA	26
Manolakos et al. 2010[150]	DD, ID	CMA	82
Muscarella et al. 2010[151]	ASD	Sanger sequencing, biochemical assay	862
Schaefer et al. 2010[152]	ASD	CMA	89
Shen et al. 2010[153]	ASD	CMA	933
Siggberg et al. 2010[154]	ID	CMA	150
Xiang et al. 2010[155]	ID	CMA	1,499
Auber et al. 2009[156]	DD, ID	Quantitative PCR	296

Table 8. Case series reporting diagnostic yield (continued)

Reference	DD Disorder	Test Studied	Sample Size
Baris and Battaloglu 2009[157]	Rett	Multiplex amplification refractory mutation system-PCR	14
Bhowmik et al. 2009[158]	FX	Methylation sensitive-PCR, biochemical assay	157
Bruno et al. 2009[42]	ID	CMA	117
Bucan et al. 2009[159]	ASD	CMA	4,310
Cho et al. 2009[160]	ID, AS, PW, VS, WS	FISH, MLPA	12
Coffee et al. 2009[61]	FX	Real-time, methylation-sensitive PCR, Southern blot	36,124
Cusco et al. 2009[161]	ASD	CMA	96
Dutta et al. 2009[162]	DD, ID, FX, congenital malformations	PCR, methylation analysis, cytogenetic analysis	179
Friedman et al. 2009[163]	ID	CMA	300
Gijsbers et al. 2009[164]	ID, multiple congenital anomalies	CMA	318
Giorda et al. 2009[165]	ID	CMA	2,400
Hochstenbach et al. 2009[166]	DD, ID	CMA	36,325
Hu et al. 2009[62]	FX	Next-generation sequencing, multiplex PCR	24
Koolen et al. 2009[167]	ID	CMA	1,364
McMullan et al. 2009[168]	ID	CMA	120
Shahdadpuri et al. 2009[169]	DD	Targeted DNA testing depending on presentation, often subtelomeric chromosome analysis	119
Utine et al. 2009[170]	ID	FISH	130
Mandal et al. 2009[171]	ID	MLPA	65
Truong et al. 2008[64]	SMS	Real-time, quantitative PCR, MLPA, FISH	64
Ballif et al. 2007[65]	DD, congenital anomalies	CMA	6,946
Baris et al. 2007[172]	DD, ID, facial dysmorphism, other congenital anomalies	CMA	373
de Souza et al. 2007[173]	DD, ID, WS	FISH	18
Newman et al. 2007[174]	DD	CMA	46
Sandrin-Garcia et al. 2007[175]	VS	FISH	16
Shen et al. 2007[66]	DD, ID, ASD, AS, PW, VS, WS	CMA	316
van Hagen et al. 2007[176]	WS	FISH, MLPA	63
Rauch et al. 2006[18]	DD, ID	CMA, FISH	1,170
Altug-Teber et al. 2005[67]	AS, PW	CMA	6
Coupry et al. 2004[177]	RTS	Real-time, quantitative PCR, microsatellite analysis	22
Kleefstra et al. 2004[178]	ID, AS, PW	Sequencing, PCR	253

AS=Angelman's syndrome (happy puppet syndrome); ASD=autism spectrum disorder (autism, autism susceptibility, MRD1, 2q23.1 deletion syndrome, 2q23.1 duplication syndrome); CMA=chromosomal microarray analysis; DD=developmental delay; FISH=fluorescence in situ hybridization; FX=fragile X (*FMR1*-related disorders, mental retardation associated with marXq28, marker X syndrome, Martin-Bell syndrome, X-linked mental retardation and macroorchidism); ID=intellectual disability; MLPA=multiplex ligation-dependent probe amplification; PCR=polymerase chain reaction; PW=Prader-Willi syndrome (Prader Labhart Willi syndrome); Rett=Rett syndrome (autism-dementia-ataxia-loss of purposeful hand use syndrome, *MECP2*-related disorders); RTS=Rubinstein-Taybi syndrome (broad thumb-hallux syndrome); SMS=Smith-Magenis syndrome (17p- syndrome, 17p11.2 monosomy, chromosome 17p11.2 deletion syndrome, chromosome 17p deletion syndrome, deletion 17p syndrome,

partial monosomy 17p); VS=velocardiofacial syndrome (22q11.2, conotruncal anomaly, DiGeorge, Shprintzen); WS=Williams syndrome (Beuren syndrome, chromosome 7q11.23 deletion syndrome, elfin facies syndrome, supravalvar aortic stenosis syndrome, Williams-Beuren syndrome)

Table 9. Case series reporting diagnostic yield by developmental disorders

DD Disorder	Number of Studies	Reference
Angelman's syndrome	7	Ahn et al. 2013[97], Halder et al. 2013[106], Ellison et al. 2012[40], Cho et al. 2009[160], Shen et al. 2007[66], Altug-Teber et al. 2005[67], Kleefstra et al. 2004[178]
Autism spectrum disorder	35	Yang et al. 2014[69], Tao et al. 2014[71], Soden et al 2014[72], Srivastava et al. 2014[47], Huguet et al. 2014[84], Stobbe et al. 2014[89], Chong et al. 2014[78], Iglesias et al. 2014[41], Koshimizu et al. 2013[53], Nicholl et al. 2014[87], Battaglia et al. 2013[98], Esposito et al. 2013[103], Fan et al. 2013[105], Marano et al. 2013[111], Nicholl et al. 2013[113], Sorte et al. 2013[120], Winarni et al. 2013[125], McGrew et al. 2012[131], Bremer et al. 2011[138], Roesser 2011[143], Muscarella et al. 2010[151], Qiao et al. 2013[116], Vallespin et al. 2013[123], Vorsanova et al. 2013[124], Zarate et al. 2013[126], Iourov et al. 2012[130], Tzetis et al. 2012[136], Bruno et al. 2011[139], Coulter et al. 2011[23], Ezugha et al. 2010[148], Schaefer et al. 2010[152], Shen et al. 2010[153], Bucan et al. 2009[159], Cusco et al. 2009[161], Shen et al. 2007[66]
Developmental delay	50	Lee et al. 2014[68], Bartnik et al. 2014[73], Boggula et al. 2014[74], Byeon et al. 2014[75], Chong et al. 2014[78], Iglesias et al. 2014[41], Pereira et al. 2014[70], Nicholl et al. 2014[87], Soden et al 2014[72], Srivastava et al. 2014[47], Tao et al. 2014[71], Uwineza et al. 2014[93], Yang et al. 2014[69], Battaglia et al. 2013[98], Doherty et al. 2013[102], Hayes et al. 2013[50], Wiszniewska et al. 2014[96], Esposito et al. 2013[103], Fan et al. 2013[105], Kashevarova et al. 2013[109], Lee et al. 2013[110], Marano et al. 2013[111], Nicholl et al. 2013[113], Pohovski et al. 2013[114], Qiao et al. 2013[116], Rodriguez-Revenga et al. 2013[117], Shoukier et al. 2013[119], Vallespin et al. 2013[122], Zarate et al. 2013[126], Dos Santos and Freire-Maia 2012[128], Ellison et al. 2012[40], Iourov et al. 2012[130], Tzetis et al. 2012[136], Bruno et al. 2011[139], Coulter et al. 2011[23], Wincent et al. 2011[146], Bernardini et al. 2010[58], Ezugha et al. 2010[148], Gervasini et al. 2010[149], Manolakos et al. 2010[150], Auber et al. 2009[156], Dutta et al. 2009[162], Hochstenbach et al. 2009[166], Shahdadpuri et al. 2009[169], Ballif et al. 2007[65], Baris et al. 2007[172], de Souza et al. 2007[173], Newman et al. 2007[174], Shen et al. 2007[66], Rauch et al. 2006[18]

Table 9. Case series reporting diagnostic yield by developmental disorders (continued)

DD Disorder	Number of Studies	Reference
Fragile X syndrome	14	Chaudhary et al. 2014[76], Fatima et al. 2014[81], Doherty et al. 2013[102], Esposito et al. 2013[103], Essop and Krause 2013[104], Jorge et al. 2013[108], Zarate et al. 2013[126], Rooms et al. 2011[144], Dave et al. 2010[147], Filipovic-Sadic et al. 2010[60], Bhowmik et al. 2009[158], Coffee et al. 2009[61], Dutta et al. 2009[162], Hu et al. 2009[62]
Intellectual disability	85	Pereira et al. 2014[70], Bartnik et al. 2014[73], Boggula et al. 2014[74], Byeon et al. 2014[75], Chaudhary et al. 2014[76], Cheon et al. 2014[77], Chong et al. 2014[78], Coutton et al. 2014[79], Fatima et al. 2014[81], Gilissen et al. 2014[82], Helsmoortel et al. 2014[83], Iglesias et al. 2014[41], Medina et al. 2014[86], Nicholl et al. 2014[87], Palmer et al. 2014[88], Pereira et al. 2014[70], Soden et al 2014[72], Tao et al. 2014[71], Tucker et al. 2014[90], Utine et al. 2014[92], Uwineza et al. 2014[93], Vergult et al. 2014[94], Willemsen and Kleefstra 2014[95], Wiszniewska et al. 2014[96], Yang et al. 2014[69], Behjati et al. 2013[99], Behjati et al. 2013[100], Jain et al. 2013[107], Tos et al. 2013[121], Hochstenbach et al. 2012[129], Rafati et al. 2012[132], Rafati et al. 2012[133], Utine et al. 2012[137], Hannibal et al. 2011[140], Rana et al. 2011[142], Shawky et al. 2011[145], Siggberg et al. 2010[154], Xiang et al. 2010[155], Bruno et al. 2009[42], Esposito et al. 2013[103], Essop and Krause 2013[104], Jorge et al. 2013[108], Kashevarova et al. 2013[109], Lee et al. 2013[110], Marano et al. 2013[111], Mundhofir et al. 2013[112], Nicholl et al. 2013[113], Pohovski et al. 2013[114], Qiao et al. 2013[116], Rodriguez-Revenga et al. 2013[117], Shoukier et al. 2013[119], Vallespin et al. 2013[122], Vallespin et al. 2013[123], Vorsanova et al. 2013[124], Zarate et al. 2013[126], Aradhya et al. 2012[127], Dos Santos and Freire-Maia 2012[128], Ellison et al. 2012[40], Iourov et al. 2012[130], Tos et al. 2012[135], Tzetis et al. 2012[136], Bruno et al. 2011[139], Coulter et al. 2011[23], Hayashi et al. 2011[141], Rooms et al. 2011[144], Bernardini et al. 2010[58], Dave et al. 2010[147], Ezugha et al. 2010[148], Manolakos et al. 2010[150], Auber et al. 2009[156], Cho et al. 2009[160], Dutta et al. 2009[162], Friedman et al. 2009[163], Gijsbers et al. 2009[164], Giorda et al. 2009[165], Hochstenbach et al. 2009[166], Koolen et al. 2009[167], McMullan et al. 2009[168], Utine et al. 2009[170], Mandal et al. 2009[171], Baris et al. 2007[172], de Souza et al. 2007[173], Shen et al. 2007[66], Rauch et al. 2006[18], Kleefstra et al. 2004[178]
Prader-Willi syndrome	9	Boggula et al. 2014[74], Kleefstra et al. 2004[178], Tuysuz et al. 2014[91], Ahn et al. 2013[97], Halder et al. 2013[106], Altug-Teber et al. 2005[67], Ellison et al. 2012[40], Cho et al. 2009[160], Shen et al. 2007[66]
Rett Syndrome	4	Kalman et al. 2014[48], Pratt et al. 2013[115], Splendore et al. 2012[134], Baris and Battaloglu 2009[157]
Smith-Magenis syndrome	4	Ahn et al. 2013[97], Dubourg et al. 2014[80], Ellison et al. 2012[40], Truong et al. 2008[64]
Rubinstein-Taybi syndrome	3	Lee et al. 2014[85], Gervasini et al. 2010[149], Coupry et al. 2004[177]
Velocardiofacial syndrome	5	Del Carmen et al. 2013[101], Ellison et al. 2012[40], Cho et al. 2009[160], Sandrin-Garcia et al. 2007[175], Shen et al. 2007[66]
Williams syndrome	6	Ellison et al. 2012[40], Cho et al. 2009[160], de Souza et al. 2007[173], Shen et al. 2007[66], van Hagen et al. 2007[176], Saad et al. 2013[118]

Studies Addressing Economic, Ethical, Social, and Legal Issues (Guiding Question 4)

For this Technical Brief, we searched for studies that addressed economic issues including cost-effectiveness of genetic testing for DDs. Our search did not identify any economic study conducted in the U.S. context. We identified several cost-effectiveness analyses conducted in other countries. We excluded these studies as the findings are not applicable to the United States because of the significant differences in the countries' economic and health care systems.

We did not identify any empirical study focusing on ethical or legal issues regarding genetic testing in the context of DD care. Limited discussions about ethical concerns (e.g., how to deal with genetic abnormalities unrelated to DDs that are detected in genome-wide testing) may exist in narrative reviews and clinical studies.[31] However, we did not include studies that provide only general discussions of ethical issues that may apply to DDs and non-DDs.

Clinical Guidelines (Guiding Question 4)

For this Technical Brief, we searched for clinical practice guidelines relevant to genetic testing for DDs that were published by medical groups or professional societies. We identified 16 relevant guidelines.[3,4,19,21,27,179-189] Table 10 is a summary of the seven guidelines that provide recommendations regarding use of genetic testing for evaluating DDs. Because genetic research and testing methods for DDs change rapidly, we did not include guidelines published beyond the last 5 years. Guidelines published before this period may be useful for some readers who are interested in certain DDs or genetic tests. Readers should always use their own judgment to determine the relevance of older guidelines to the topics of their interest. Guidelines that focus only on interpreting genetic testing results are also not included in Table 10. Because of the differences in their purposes and methodologies, these guidelines provide different recommendations. ASD or ID are addressed in six of the seven guidelines. CMA testing for CNV is recommended for use in evaluating individuals with ASD, ID, GDD, or certain congenital anomalies in four guidelines.[3,19,21,27] See Table 10 for detailed recommendations.

Emerging Technologies and Ongoing Trials (Guiding Question 4)

The GTR data we collected did not allow an analysis to predict which type of genetic tests will be more prevalent in DD care in the future. We did not identify any data-based analysis that predicted the trend of genetic technologies for DD diagnosis or screening. However, our interview of the Key Informants suggested that the whole exome or genome sequencing may be increasingly used in the context of DD care as the cost for these tests continue to drop. See the section on whole-exome and whole-genome sequencing in Appendix B for more information on these technologies and their technical capabilities. Additional references are provided in that section for a more in-depth overview of the sequencing technologies, including their potential advantages and limitations.

Our search of the National Clinical Trials online database (www.ClinicalTrials.gov) identified 10 ongoing clinical trials. The purposes of these trials vary significantly. Six trials are intended to explore the genetic mechanisms or genotype-phenotype association for ASD (ClinicalTrials.gov identifiers NCT01686685, NCT01749670, NCT01646866, NCT01770548) or ID (NCT01867554, NCT02136849). Two trials are purported to validate the algorithm (NCT01810341) or the sample collecting method (NCT01616589) used in a genetic test.

Another two trials are intended to study the effectiveness of a treatment in patients selected based on genotyping findings (NCT00768820, NCT00859664). Eight of the 10 trials focus on ADD or ID. Seven of the 10 trials are observational cohort studies. The other three trials include two case-control studies and a nonrandomized, parallel assignment study. These trials are summarized in Appendix E.

Table 10. Summary of recent clinical guidelines

Reference	Purpose	Disorders Addressed	Recommendation Relevant to Genetic Testing
Moeschler et al. 2014[3] American Academy of Pediatrics Committee on Genetics	ID or GDD	To describe an optimal medical genetics evaluation of the child with ID or GDD. This report does not cover children with ASD who also have ID as a co-occurring disability or children with a single-domain developmental delay.	• If a specific diagnosis is suspected, arrange for the appropriate diagnostic studies to confirm including single-gene tests or chromosomal microarray test. • If diagnosis is unknown and no clinical diagnosis is strongly suspected, begin the stepwise evaluation process: a. CMA should be performed in all. b. metabolic testing should be considered and should include serum total homocysteine, acyl-carnitine profile, amino acids; and urine organic acids, glycosaminoglycans, oligosaccharides, purines, pyrimidines, guanidinoacetate /creatine metabolites. c. Fragile X testing should be performed in all. • If no diagnosis is established: a. Male gender and family history suggestive X-linkage, complete XLID panel that contains genes causal of nonsyndromic XLID and complete high density X-CMA. Consider X-inactivation skewing in the mother of the proband. b. Female gender: complete *MECP2* deletion, duplication, and sequencing study. If the specific diagnosis is certain, provide genetic counseling services by a certified genetic counselor.

Table 10. Summary of recent clinical guidelines (continued)

Reference	Purpose	Disorders Addressed	Recommendation Relevant to Genetic Testing
Schaefer et al. 2013[19] American College of Medical Genetics and Genomics	ASD	To present a tiered evaluation approach of the etiology of ASD based on current evidence to assist clinicians	• A genetic evaluation should be offered to every person with ASD. • In situations in which 3rd-party payers will cover cytogenetic studies but not CMA testing, a conventional chromosomal analysis is preferable to no cytogenetic testing at all. • Because 1 ASD hotspot (16p11.2) has been reported to have CNVs occurring in 0.5% to 1% of all individuals with ASD, CMA is now recommended as a 1st-tier test over karyotyping. • There is adequate evidence to suggest testing for fragile X syndrome, methyl-CPG-binding protein 2 spectrum disorders, and PTEN-related conditions in patients with ASD with no other identifiable etiology. • Routine testing of females with ASD for fragile X does not meet evidence-based criteria. However, serious consideration should be given to order fragile X testing in females with ASD when prompted by clinical parameters such as a phenotype compatible with fragile X, a family history positive for X-linked neurodevelopmental disorders, or premature ovarian insufficiency, ataxia, or tremors in close relatives. • Given the current evidence, *MECP2* testing of males with autism is not recommended. However, geneticists should be alert to the features of *MECP2* duplications (drooling, recurrent respiratory infections, hypotonic facies) and consider *MECP2* duplication testing in boys with autism and such features. • It is suggested that PTEN testing be reserved for patients with ASD with a head circumference above the 98th percentile. When a family history is consistent with X linked inheritance and the patient has cognitive impairments, an X-linked intellectual disability gene panel is a consideration. • Testing for mitochondrial disorders in persons with ASD is recommended only if supporting symptoms or laboratory abnormalities are present. • Genetic tests that have been suggested in the etiologic evaluation of ASD but currently with insufficient evidence to recommend routine testing include: *CDLK5* testing, cholesterol/7 dehydrocholesterol, chromosome 15 methylation/*UBE3A* gene testing, methylation/epigenetic testing, mitochondrial gene sequencing/oligoarray, *NSD1* testing, reduction-oxidation studies, purine/pyrimidine metabolism, folate-sensitive fragile sites, and selected neurometabolic screening.

Table 10. Summary of recent clinical guidelines (continued)

Reference	Purpose	Disorders Addressed	Recommendation Relevant to Genetic Testing
Finucane et al. 2012[190] National Society of Genetic Counselors	*FMR1*-associated disorders	To assist genetic counselors in providing accurate risk assessment and appropriate educational and supportive counseling for individuals with positive test results and families affected by *FMR1*-associated disorders	No specific genetic test was recommended by this guidelines authors.
National Institute for Health and Care Excellence (UK) 2011[181]	ASD	To provide information on the recognition, referral, and diagnosis of autism in children and young people from birth through 19 years of age	• Consider whether the child or young person may have medical or genetic problems and disorder (e.g., chromosome disorders, genetic abnormalities including fragile X) as a coexisting condition and, if suspected, carry out appropriate assessments and referrals. • Do not routinely perform any medical investigations as part of an autism diagnostic assessment but consider the following in individual circumstances and based on physical examination, clinical judgment and the child or young person's profile: genetic tests, as recommended by your regional genetics center, if there are specific dysmorphic features, congenital anomalies and/or evidence of intellectual disability.
Manning et al. 2010[27] American College of Medical Genetics and Genomics	ID, ASD, GDD, or congenital anomalies	To provide guidance for health care providers treating patients with developmental delays, ID, congenital anomalies, dysmorphic features, and ASD in determining the need for array-based genetic testing for detecting chromosomal abnormalities	• CMA testing for CNVs is recommended as a 1st-line test in the initial postnatal evaluation of individuals with the following: o Multiple anomalies not specific to a well-delineated genetic syndrome o Apparently nonsyndromic developmental delays or intellectual disabilities o ASD • Appropriate followup is recommended in cases of chromosome imbalance identified by CMA, to include cytogenetic/FISH studies of the patient, parental evaluation, and clinical genetic evaluation and counseling.
Miller et al. 2010[21] International Standard Cytogenomic Array Consortium	ID, ASD, or multiple congenital anomalies	To evaluate the benefits and limitations of CMA as compared with G-banded karyotyping for detecting pathogenic genomic imbalances in patients with ID, ASD, and/or multiple congenital anomalies	The authors recommended offering CMA as the 1st-tier genetic test, in place of G-banded karyotype, for patients with unexplained developmental delays/ID, ASD, or multiple congenital anomalies.

Table 10. Summary of recent clinical guidelines (continued)

Reference	Purpose	Disorders Addressed	Recommendation Relevant to Genetic Testing
Ministry of Health, Singapore 2010[182]	ASD	To assist practitioners in Singapore who are involved in any of the following: surveillance, screening, and early identification, referral for assessment, diagnosis and intervention of children with ASD	• Children with ASD with the following features should have a genetic evaluation: microencephaly or macroencephaly, a positive family history of a genetic syndrome, dysmorphic features. • Children with ASD may be offered high-resolution chromosomal studies and DNA analysis to look for an associated medical condition after diagnosis.

ASD=autism spectrum disorder; CMA= chromosomal microarray analysis; CNV= copy-number variation; FISH=fluorescent in situ hybridization; GDD=global developmental delay; ID=intellectual disability; PTEN= phosphate and tensin homolog; XLID= X-linked intellectual disability

Systematic Reviews and Technology Assessment Reports (Guiding Question 4)

We searched for systematic reviews and health technology assessment reports relevant to genetic testing for DD, ID, and ASD that were published by professional societies in the past 5 years. Our search identified three relevant documents. These systematic reviews addressed diagnostic yields of microarray-based comparative genomic hybridization (aCGH), testing for X-linked ID genes, *FMR1* testing, *MeCP2* testing, and conventional G-banded karyotyping for developmental delays, ID, or ASD. The main findings of these reviews are summarized in Table 11. We did not include systematic reviews or technology assessment reports published in non-English languages. We also excluded proprietary technology assessment reports that are not publicly accessible. Because genetic research and testing methods for DDs change rapidly, we did not search for systematic reviews or technology assessment reports published beyond the most recent 5 years.

Table 11. Summary of recent systematic reviews and technology assessment reports

Reference	Purpose	Resources Searched and Inclusion Criteria	Findings
Hochstenbach et al. 2011[191]	To review the contributions and limitations of genome-wide array-based identification of CNVs in the clinical diagnostic evaluation of patients with MR and other brain-related disorders	Publications were retrieved from the PubMed database of the National Center for Biotechnology Information. The studies were included only if (1) the clinical diagnosis was made according to international standards, (2) segmental aneuploidies detected by array-based methods was validated by an independent method, or (3) it was possible to relate the aberrations to specific patients.	In unselected MR referrals, a causative genomic gain or loss is detected in 14% to18% of cases. Usually, such CNVs arise de novo, are not found in healthy subjects, and have a major impact on the phenotype by altering the dosage of multiple genes. The expected diagnostic yield for patients with autism is about 5% to 10% in nonsyndromic and 10% to 20% in syndromic patients. Exome sequencing in patients with MR or autism revealed de novo mutations in protein coding genes in 60% and 20% of cases, respectively.

Table 11. Summary of recent systematic reviews and technology assessment reports (continued)

Reference	Purpose	Resources Searched and Inclusion Criteria	Findings
Michelson et al. 2011[20]	To systematically review the evidence concerning the diagnostic yield of genetic and metabolic evaluation of children with GDD/ID	Relevant literature was reviewed, abstracted, and classified according to the 4-tiered American Academy of Neurology classification of evidence scheme	In patients with GDD/ID, microarray testing is diagnostic on average in 7.8%, G-banded karyotyping is abnormal in at least 4%, and subtelomeric fluorescence in situ hybridization is positive in 3.5%. Testing for X-linked ID genes has a yield of up to 42% in males with an appropriate family history. *FMR1* testing shows full expansion in at least 2% of patients with mild to moderate GDD/ID, and *MeCP2* testing is diagnostic in 1.5% of females with moderate to severe GDD/ID.
Sagoo et al. 2009[192]	To update a previous systematic review evaluating array-based comparative genomic hybridization used in patients with ID and congenital anomalies	MEDLINE, EMBASE, and Web of Science databases were searched during March 2008 with both free-text and MeSH terms. No language or other search restrictions were imposed and reference lists of primary studies were checked for additional references.	The overall diagnostic yield of causal abnormalities was 10%. The overall number needed to test to identify an extra causal abnormality was 10. The overall false-positive yield of noncausal abnormalities was 7%.

CNVs=copy number variation; GDD=global developmental delay; ID=intellectual disability; MR=mental retardation

Summary and Implications

Scientific advances in recent decades have led to the discovery of genetic abnormalities that may explain the reasons for many developmental disability (DD) cases. A large number of genetic tests have been developed and adopted in clinical practice. These tests are used to differentiate well-defined DD syndromes (e.g., fragile X syndrome, Rett syndrome) or, more commonly, to establish an etiologic diagnosis for unexplained intellectual disability (ID), autism spectrum disorder (ASD), or global developmental delay (GDD). For this Technical Brief, we identified 672 genetic tests for 11 DD categories using the National Center for Biotechnology Information's Genetic Testing Registry (GTR) database. These tests employ a broad range of methods, including next-generation sequencing, Sanger sequence analysis, microarray, comparative genomic hybridization, single nucleotide polymorphism detection, multiplex ligation-dependent probe amplification, and other polymerase chain reaction–based tests. These tests analyze a single gene, a chromosome, a chromosomal region, or the whole genome or exome.

Our search identified a single U.S. Food and Drug Administration (FDA)-cleared commercial test kit. All other 672 tests are laboratory-developed tests (LDTs). These LDTs are offered by 63 laboratories certified under the Clinical Laboratory Improvement Amendments of 1988 in 29 States. Patients in some States may not have access to certain LDTs. FDA does not actively regulate LDTs at this time; however, on October 3, 2014, FDA published two draft guidance documents regarding oversight of LDTs. The proposed policy change has some significant implications for the LDTs we compiled in this Technical Brief. When The FDA guidance documents are finalized, we will have a better chance to evaluate how LDTs will be regulated in the future.

As genetic tests have become increasingly available, payers have observed a rapid diffusion of these tests in health care. Some tests (e.g., microarray-based comparative genomic hybridization [aCGH]) have been recommended by professional groups as first-tier diagnostic tests for DDs. The proposed benefits of genetic testing include providing an improved sense of empowerment for patient families, refining treatment options, providing prognosis, preventing comorbidities, avoiding unnecessary diagnostic tests, providing recurrence-risk-based counseling, and improving access to needed support or services. However, these proposed benefits need to be validated by clinical studies.

One major goal of this Technical Brief is to identify existing evidence for addressing the clinical utility of genetic tests for DDs. To achieve the goal, we thoroughly scanned medical literature (see Table 12 below for a summary of this effort). We focused on evidence directly linking genetic testing to changes in health outcomes. However, our search did not identify any study—randomized or nonrandomized—in that category. We consider this a major gap that needs to be filled by future research. Randomized controlled trials (RCTs) and well-designed nonrandomized studies that directly compare health outcomes for use versus no use of the tests is the ideal type of study for addressing clinical utility. However, conducting these studies, particularly RCTs, can be difficult for various practical reasons. Because the genetic testing area changes so quickly, the test being studied may become obsolete even before long-term data are available. Other practical challenges for conducting clinical utility trials also exist, such as difficulty in patient recruitment (particularly for rare disorders) and high expense associated with the studies. Regardless of these challenges, it may still be feasible to design and execute clinical utility trials for certain tests and disorders, and we encourage researchers to make an effort in that direction.

Besides searching clinical trials, we also searched for other types of evidence that may contribute to establishing an indirect linkage between genetic testing and health outcomes. We identified a small number of studies assessing genetic tests' value perceived by families affected by DDs or addressing the impact of genetic testing on clinical management or family decisions. This type of study enhances our understanding of genetic tests' potential to cause changes in health outcomes (e.g., psychosocial outcomes). For example, one survey we identified reported that some parents of children with DD considered a clinical diagnosis (e.g., autism) more useful a label than a rare, specific etiological diagnosis.[43] The intensity of their need for an etiologic diagnosis was reported to diminish over time. A few case series we identified reported changes being made in patient management due to the findings of genetic tests. More studies should be performed to investigate these important issues, particularly parents' views on the importance of determining etiology and how to counsel them on the value of etiologic evaluation.

Given that most of the genetic tests relevant to this report are rarely used in isolation to establish a clinical diagnosis, we only identified a single case-control study that addressed clinical validity (i.e., diagnostic accuracy). Our literature search identified 20 studies addressing analytic validity issues. Most of these studies validated the analytic performance of a newly developed test, reporting on the test's sensitivity, specificity, predictive values, concordance, or repeatability. The findings of these studies of new tests need to be further validated in future research.

We identified a large number (129) of case series that reported on the diagnostic yield of a genetic test. These case series constitute the largest portion of the evidence base we report in this Technical Brief. However, no consensus has been reached on the usefulness of diagnostic yield studies in assessing a genetic test's clinical utility (impact on health outcomes). Although diagnostic yield indicates the percentage of patients being tested who ultimately reach a diagnosis, it does not confirm whether the diagnoses reached are correct. Improved diagnostic yield may have an impact on patient management because some condition-specific management (e.g., obesity prevention programs for DD patients at higher risk for developing the comorbidity) cannot be initiated without a specific diagnosis (e.g., an etiologic diagnosis revealing a patient carries a genetic variant associated with obesity). However, patient management changes based on the testing may not necessarily lead to improved health outcomes.

In the context of DD care, genetic testing is often used to establish an etiologic diagnosis rather than establish a clinical diagnosis. However, researchers may not always agree on whether a genetic aberration (e.g., certain type of copy number variants) is "causal," "pathogenic," or "clinically significant." Several public information sources exist to facilitate the identification of causal genetic aberrations. These sources include the National Center for Biotechnology Information; the International Standards for Cytogenomic Arrays; American College of Medical Genetics and Genomics Practice Guidelines; the University of California, Santa Cruz, Genome Browser; the Database of Genomic Variants; and the Genoglyphix Chromosome Aberration Database. However, the existence of these databases does not completely eliminate the uncertainty in certain genetic aberrations' causal role in DDs. A more robust framework for evaluating which variants play a role in disease and are relevant to patient care is needed.[193] The uncertainty in the current databases discounts some diagnostic-yield studies' validity. Ongoing efforts, such as the National Institutes of Health–funded Clinical Genome Resource (ClinGen), may provide valuable information for identifying clinically relevant genetic variants.[193]

For this Technical Brief, we also searched for studies that addressed cost, ethical, legal, and social issues related to using genetic tests for DDs. However, we did not identify any economic

studies performed in the U.S. context. We did not identify any empirical research on legal, and social issues specifically related to genetic testing for DDs. As a result, the impact of genetic testing in those areas remains unclear.

Table 12 below summarizes the evidence gaps we previously discussed. We referred to several guidance documents published by the Agency for Healthcare Research and Quality and the Cochrane Library to determine the types of evidence required for addressing a genetic test's clinical utility.[36,194,195] As the table indicates, evidence that directly or indirectly supports genetic testing's clinical utility is generally thin. Significant investment in research to fill the gaps may be needed. Additionally, the effectiveness of genetic testing (e.g., its accuracy, impact on health outcomes) may be affected by who orders the tests or who interprets the test results. Our literature search did not identify any comparison study that address this concern, which is another gap that needs to be filled by future research.

This Technical Brief has several limitations. First, we primarily relied on GTR to identify genetic tests. Because the GTR data were voluntarily submitted by test providers, genetic tests that were not reported to GTR are not captured by this brief. Although the GTR database is arguably one of the most comprehensive sources on genetic tests, how well the tests we identified represent the whole landscape of genetic testing for DDs is not clear.

Second, within the timeframe of this Technical Brief, we were not able to independently verify the accuracy of the GTR data submitted by test providers. Although the GTR implements a solid data quality assurance program,[38] we are not certain about the program's effectiveness. Readers of this report should use caution when they interpret the GTR data we collected.

Third, DDs include a large number of disorders. This Technical Brief focuses only on some common DD conditions or syndromes based on the need of the key stakeholders for this project. Genetic tests for many rare DD syndromes are not within the scope of the brief. The findings of this Technical Brief should be interpreted as only applying to the DD conditions included in the report.

Fourth, this Technical Brief is intended to collect basic information on genetic tests for DDs. It is not a systematic review. Although we performed a systematic search for evidence that potentially addresses genetic tests' clinical utility, we did not comprehensively evaluate the strength of evidence. To have a more in-depth understanding of how well the evidence has addressed the clinical utility issues, a series of systematic reviews may be needed.

Despite these limitations, this Technical Brief provides useful information for understanding the current landscape of genetic testing for DDs. The evidence gaps identified will help guide future research to generate the most needed evidence for addressing genetic tests' clinical utility in the context of DD care.

Table 12. Evidence gap

Domain	Ideal Evidence	Helpful Evidence	Number of Studies Identified
Clinical utility	RCTs directly evaluating if use of the test affects clinical outcomes		0
Clinical utility		Non-RCT studies directly evaluating whether use of the test affects clinical outcomes	0
Clinical utility		Studies that evaluate the impact of testing on clinical management or family decisions	8
Clinical utility		Studies that evaluate patients or families' preference for genetic testing	2
Clinical utility		Diagnostic case series	129
Clinical validity*	Cohort studies that evaluate the test's diagnostic accuracy using phenotype or other acceptable diagnostic standards as reference methods		0
Clinical validity*		Case-control studies that evaluate the test's diagnostic accuracy	1
Analytic validity	Case-control studies using appropriately validated samples to evaluate the test's analytic accuracy		15
Analytic validity		Case series that study analytic performance	5
Analytic validity	Studies reporting on the findings of external proficiency testing programs		0
Analytic validity	Bench studies that evaluate a test's repeatability, reproducibility, and other performance characteristics		0
Economic issue	Cost-effectiveness analysis		0
Economic analysis		Economic-impact studies, cost reports	0
Ethical, legal, social impact	Surveys; reports of consensus-based opinions		0

*Given that most of the genetic tests relevant to this report are rarely used in isolation to establish a clinical diagnosis, the number of studies that report clinical validity (i.e., diagnostic accuracy) is expected to be small.

RCT=randomized controlled trial

References

1. National Center on Birth Defects and Developmental Disabilities (NCBDDD). Facts about developmental disabilities. Atlanta (GA): Centers for Disease Control and Prevention (CDC); 2013 Dec 26. http://www.cdc.gov/ncbddd/developmentaldisabilities/facts.html. Accessed 2014 Apr 01.
2. Boyle CA, Boulet S, Schieve LA, et al. Trends in the prevalence of developmental disabilities in US children, 1997-2008. Pediatrics. 2011 Jun;127(6):1034-42. PMID: 21606152.
3. Moeschler JB, Shevell M, Committee on Genetics. Comprehensive evaluation of the child with intellectual disability or global developmental delays. Pediatrics. 2014 Sep;134(3):e903-18. PMID: 25157020.
4. Shevell M, Ashwal S, Donley D,et al. Practice parameter: evaluation of the child with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and The Practice Committee of the Child Neurology Society. Neurology. 2003 Feb 11;60(3):367-80. PMID: 12578916.
5. Roeleveld N, Zielhuis GA, Gabreels F. The prevalence of mental retardation: a critical review of recent literature. Dev Med Child Neurol. 1997 Feb;39(2):125-32.
6. Schalock RL, Luckasson RA, Shogren KA, et al. The renaming of mental retardation: understanding the change to the term intellectual disability. Intellect Dev Disabil. 2007 Apr;45(2):116-24. PMID: 17428134.
7. Frequently asked questions on intellectual disability. Washington (DC): American Association on Intellectual and Developmental Disabilities (AAIDD). http://aaidd.org/intellectual-disability/definition/faqs-on-intellectual-disability#.VCBG9Xl0wUR. Accessed 2014 Sep 22.
8. What causes IDDs? Bethesda (MD): Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD); 2012 Nov 30. http://www.nichd.nih.gov/health/topics/idds/conditioninfo/Pages/causes.aspx. Accessed 2014 Sep 22.
9. Intellectual disability. Elk Grove Village (IL): American Academy of Pediatrics; 2014 Sep 02. http://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/pages/Intellectual-Disability.aspx. Accessed 2014 Sep 22.
10. Developmental Disabilities Monitoring Network Surveillance Year 2010 principal investigators, Centers for Disease Control and Prevention (CDC). Prevalence of autism spectrum disorder among children aged 8 years - autism and developmental disabilities monitoring network, 11 sites, United States, 2010. Morb Mortal Wkly Rep Surveill Summ. 2014 Mar 28;63(2):1-21. PMID: 24670961.
11. Freitag CM, Staal W, Klauck SM, et al. Genetics of autistic disorders: review and clinical implications. Eur Child Adolesc Psychiatry. 2010 Mar;19(3):169-78. PMID: 19941018.
12. State MW. The genetics of child psychiatric disorders: focus on autism and Tourette syndrome. Neuron. 2010 Oct 21;68(2):254-69. PMID: 20955933.
13. Volkmar F, Siegel M, Woodbury-Smith M, et al. Practice parameter for the assessment and treatment of children and adolescents with autism spectrum disorder. J Am Acad Child Adolesc Psychiatry. 2014 Feb;53(2):237-57. PMID: 24472258.
14. Moeschler JB. Medical genetics diagnostic evaluation of the child with global developmental delay or intellectual disability. Curr Opin Neurol. 2008 Apr;21(2):117-22. PMID: 18317267.
15. Shevell M, Ashwal S, Donley D, et al. Practice parameter: evaluation of the child with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society [slide set]. St. Paul (MN): American Academy of Neurology; 2003. 60 p.
16. McDonald L, Rennie A, Tolmie J, et al. Investigation of global developmental delay. Arch Dis Child. 2006 Aug;91(8):701-5. PMID: 16861488.

17. Kaufman L, Ayub M, Vincent JB. The genetic basis of non-syndromic intellectual disability: a review. J Neurodev Disord. 2010 Dec;2(4):182-209. PMID: 21124998.

18. Rauch A, Hoyer J, Guth S, et al. Diagnostic yield of various genetic approaches in patients with unexplained developmental delay or mental retardation. Am J Med Genet A. 2006 Oct 1;140(19):2063-74. PMID: 16917849.

19. Schaefer GB, Mendelsohn NJ. Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. Genet Med. 2013 May;15(5):399-407. PMID: 23519317.

20. Michelson DJ, Shevell MI, Sherr EH, et al. Evidence report: genetic and metabolic testing on children with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. Neurology. 2011 Oct 25;77(17):1629-35. PMID: 21956720.

21. Miller DT, Adam MP, Aradhya S, et al. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010 May 14;86(5):749-64. PMID: 20466091.

22. Mroch AR, Flanagan JD, Stein QP. Solving the puzzle: case examples of array comparative genomic hybridization as a tool to end the diagnostic odyssey. Curr Probl Pediatr Adolesc Health Care. 2012 Mar;42(3):74-8. PMID: 22325475.

23. Coulter ME, Miller DT, Harris DJ, et al. Chromosomal microarray testing influences medical management. Genet Med. 2011 Sep;13(9):770-6. PMID: 21716121.

24. Riggs E, Wain K, Riethmaier D, et al. Chromosomal microarray impacts clinical management. Clin Genet. 2013 Feb;85(2):147-53. Epub 2013 Feb 21. PMID: 23347240.

25. Saam J, Gudgeon J, Aston E, et al. How physicians use array comparative genomic hybridization results to guide patient management in children with developmental delay. Genet Med. 2008 Mar;10(3):181-6. PMID: 18344707.

26. Turner G, Boyle J, Partington MW, et al. Restoring reproductive confidence in families with X-linked mental retardation by finding the causal mutation. Clin Genet. 2008 Feb;73(2):188-90. PMID: 18070138.

27. Manning M, Hudgins L. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genet Med. 2010 Nov;12(11):742-5. PMID: 20962661.

28. What is the comparative effectiveness of genetic and other diagnostic tests to identify and treat intellectual and developmental disabilities caused by genetic mutations in post-natal to early childhood populations? Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2013 Oct 26. http://effectivehealthcare.ahrq.gov/submit-a-suggestion-for-research/read-suggested-topics-for-research/?pageAction=view&topicID=594&source=current. Accessed 2014 Jun 24.

29. Special report: aCGH for the genetic evaluation of patients with developmental delay/mental retardation or autism spectrum disorder (Structured abstract). Health Tech Assess Database. 2008;(1) http://onlinelibrary.wiley.com/o/cochrane/clhta/articles/HTA-32010000222/frame.html.

30. Topic triage cover sheet: Genetic testing for developmental diseases. Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2012 Mar 6. 5 p.

31. Jordan BR, Tsai DF. Whole-genome association studies for multigenic diseases: ethical dilemmas arising from commercialization--the case of genetic testing for autism. J Med Ethics. 2010 Jul;36(7):440-4. PMID: 20558435.

32. Secretary's Advisory Committee on Genetics, Health, and Society (SACGHS). U.S. system of oversight of genetic testing: a response to the charge of the Secretary of Health and Human Services. Washington (DC): Department of Health & Human Services; 2008 Apr. 276 p. http://www4.od.nih.gov/oba/SACGHS/reports/SACGHS_oversight_report.pdf.

33. Sun F, Bruening W, Uhl S, et al. Quality, regulation and clinical utility of laboratory-developed molecular tests (Prepared by ECRI Institute Evidence-based Practice Center under Contract No. 290-2007-1063 I). Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2010 May 19. 217 p. http://www.cms.gov/determinationprocess/downloads/id72TA.pdf.

34. U.S. Food and Drug Administration (FDA), Center for Biologics Evaluation and Research. Draft guidance for industry, Food and Drug Administration staff, and clinical laboratories: FDA notification and medical device reporting for laboratory developed tests (LDTs). Silver Spring (MD): U.S. Food and Drug Administration (FDA); 2014 Oct 3. 28 p. http://www.fda.gov/ucm/groups/fdagov-public/%40fdagov-meddev-gen/documents/document/ucm416684.pdf.

35. U.S. Food and Drug Administration (FDA), Center for Biologics Evaluation and Research. Draft guidance for industry, Food and Drug Administration staff, and clinical laboratories: framework for regulatory oversight of laboratory developed tests (LDTs). Silver Spring (MD): U.S. Food and Drug Administration (FDA); 2014 Oct 3. 41 p. http://www.fda.gov/downloads/MedicalDevices/DeviceRegulationandGuidance/GuidanceDocuments/UCM416685.pdf.

36. Sun F, Bruening W, Erinoff E, et al. Addressing challenges in genetic test evaluation. Evaluation frameworks and assessment of analytic validity Methods research report. (Prepared by the ECRI Institute Evidence-based Practice Center under Contract No. HHSA 290-2007-10063-I.) AHRQ publication no. 11-EHC048-EF. Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2011 Jun. 107 p. http://www.ncbi.nlm.nih.gov/books/NBK56750/pdf/TOC.pdf.

37. Sun F, Schoelles KM, Coates VH. Assessing the utility of genetic tests. J Ambul Care Manage. 2013 Jul-Sep;36(3):222-32. PMID: 23748269.

38. Rubinstein WS, Maglott DR, Lee JM, et al. The NIH genetic testing registry: a new, centralized database of genetic tests to enable access to comprehensive information and improve transparency. Nucleic Acids Res. 2013 Jan;41:D925-35. PMID: 23193275.

39. Evaluation of automatic class III designation for Affymetrix CytoScan Dx Assay decision summary. K130313. Silver Spring (MD): U.S. Food and Drug Administration; 44 p. http://www.accessdata.fda.gov/cdrh_docs/reviews/K130313.pdf.

40. Ellison JW, Ravnan JB, Rosenfeld JA, et al. Clinical utility of chromosomal microarray analysis. Pediatrics. 2012 Nov;130(5):e1085-e1095. PMID: 23071206.

41. Iglesias A, Anyane-Yeboa K, Wynn J, et al. The usefulness of whole-exome sequencing in routine clinical practice. Genet Med. 2014 Jun 5;Epub ahead of print. PMID: 24901346.

42. Bruno DL, Ganesamoorthy D, Schoumans J, et al. Detection of cryptic pathogenic copy number variations and constitutional loss of heterozygosity using high resolution SNP microarray analysis in 117 patients referred for cytogenetic analysis and impact on clinical practice. J Med Genet. 2009 Feb;46(2):123-31. PMID: 19015223.

43. Makela NL, Birch PH, Friedman JM, et al. Parental perceived value of a diagnosis for Intellectual Disability (ID): A qualitative comparison of families with and without a diagnosis for their child's ID. Am J Med Genet A. 2009 Nov;149(11):2393-402. PMID: 19842198.

44. Costain G, Chow EW, Ray PN, et al. Caregiver and adult patient perspectives on the importance of a diagnosis of 22q11.2 deletion syndrome. J Intellect Disabil Res. 2012 Jun;56(6):641-51. PMID: 22142442.

45. Amiet C, Couchon E, Carr K, et al. Are there cultural differences in parental interest in early diagnosis and genetic risk assessment for autism spectrum disorder? Front Pediatr. 2014;2:32. PMID: 24795872.

46. Jiao Y, Chen R, Ke X, et al. Predictive models for subtypes of autism spectrum disorder based on single-nucleotide polymorphisms and magnetic resonance imaging. Adv Med Sci. 2011 Dec 1;56(2):334-42. PMID: 22037176.

47. Srivastava S, Cohen JS, Vernon H, et al. Clinical whole exome sequencing in child neurology practice. Ann Neurol. 2014 Oct. http://dx.doi.org/10.1002/ana.24226.

48. Kalman LV, Tarleton JC, Percy AK, et al. Development of a genomic DNA reference material panel for Rett syndrome (MECP2-related disorders) genetic testing. J Mol Diagn. 2014 Mar;16(2):273-9. PMID: 24508304.

49. Inaba Y, Schwartz CE, Bui QM, et al. Early detection of fragile X syndrome: Applications of a novel approach for improved quantitative methylation analysis in venous blood and newborn blood spots. Clin Chem. 2014 Jul;60(7):963-73.

50. Hayes JL, Tzika A, Thygesen H, et al. Diagnosis of copy number variation by Illumina next generation sequencing is comparable in performance to oligonucleotide array comparative genomic hybridisation. Genomics. 2013 Sep;102(3):174-81. PMID: 23598253.

51. Stofanko M, Han JC, Elsea SH, et al. Rapid and inexpensive screening of genomic copy number variations using a novel quantitative fluorescent PCR method. Dis Markers. 2013;35(6):589-94. PMID: 24288428.

52. Stofanko M, Goncalves-Dornelas H, Cunha PS, et al. Simple, rapid and inexpensive quantitative fluorescent PCR method for eetection of microdeletion and microduplication syndromes. PLoS ONE. 2013 Apr 19;8(4):e61328. PMID: 23620743.

53. Koshimizu E, Miyatake S, Okamoto N, et al. Performance comparison of bench-top next generation sequencers using microdroplet PCR-based enrichment for targeted sequencing in patients with autism spectrum disorder. PLoS ONE. 2013 Sep 16;8(9):e74167.

54. Lafauci G, Adayev T, Kascsak R, et al. Fragile X screening by quantification of FMRP in dried blood spots by a luminex immunoassay. J Mol Diagn. 2013 Jul;15(4):508-17. PMID: 23660422.

55. Curtis-Cioffi KM, Rodrigueiro DA, Rodrigues VC, et al. Comparison between the polymerase chain reaction-based screening and the southern blot methods for identification of fragile X syndrome. Genet Test Mol Biomar. 2012 Nov 1;16(11):1303-8. PMID: 23101592.

56. Juusola JS, Anderson P, Sabato F, et al. Performance evaluation of two methods using commercially available reagents for PCR-based detection of FMR1 mutation. J Mol Diagn. 2012 Sep;14(5):476-86. PMID: 22765921.

57. Lessard M, Chouiali A, Drouin R, et al. Quantitative measurement of FMRP in blood platelets as a new screening test for fragile X syndrome. Clin Genet. 2012 Nov;82(5):472-7. PMID: 21992468.

58. Bernardini L, Alesi V, Loddo S, et al. High-resolution SNP arrays in mental retardation diagnostics: how much do we gain? Eur J Hum Genet. 2010 Feb;18(2):178-85. PMID: 19809473.

59. Chen L, Hadd A, Sah S, et al. An information-rich CGG repeat primed PCR that detects the full range of fragile X expanded alleles and minimizes the need for southern blot analysis. J Mol Diagn. 2010 Sep;12(5):589-600. PMID: 20616364.

60. Filipovic-Sadic S, Sah S, Chen L, et al. A novel FMR1 PCR method for the routine detection of low abundance expanded alleles and full mutations in fragile X syndrome. Clin Chem. 2010 Mar;56(3):399-408. PMID: 20056738.

61. Coffee B, Keith K, Albizua I, et al. Incidence of Fragile X Syndrome by newborn screening for methylated FMR1 DNA. Am J Hum Genet. 2009 Oct 9;85(4):503-14. PMID: 19804849.

62. Hu H, Wrogemann K, Kalscheuer V, et al. Mutation screening in 86 known X-linked mental retardation genes by droplet-based multiplex PCR and massive parallel sequencing. Hugo J. 2009 Dec;3(1-4):41-9. PMID: 21836662.

63. Wang W, Law HY, Chong SS. Detection and discrimination between deletional and non-deletional prader-willi and angelman syndromes by methylation-specific pcr and quantitative melting curve analysis. J Mol Diagn. 2009 Sep;11(5):446-9. PMID: 19661385.

64. Truong HT, Solaymani-Kohal S, Baker KR, et al. Diagnosing Smith-Magenis syndrome and duplication 17p11.2 syndrome by RAI1 gene copy number variation using quantitative real-time PCR. Genet Test. 2008 Mar;12(1):67-73. PMID: 18373405.

65. Ballif BC, Sulpizio SG, Lloyd RM, et al. The clinical utility of enhanced subtelomeric coverage in array CGH. Am J Med Genet A. 2007 Aug 15;143A(16):1850-7. PMID: 17632771.

66. Shen Y, Irons M, Miller DT, et al. Development of a focused oligonucleotide-array comparative genomic hybridization chip for clinical diagnosis of genomic imbalance. Clin Chem. 2007 Dec;53(12):2051-9. PMID: 17901113.

67. Altug-Teber O, Dufke A, Poths S, et al. A rapid microarray based whole genome analysis for detection of uniparental disomy. Hum Mutat. 2005 Aug;26(2):153-9. PMID: 15968682.

68. Lee H, Deignan JL, Dorrani N, et al. Clinical exome sequencing for genetic identification of rare mendelian disorders. JAMA. 2014 Nov 12;312(18):1880-7. http://dx.doi.org/10.1001/jama.2014.14604.

69. Yang Y, Muzny DM, Xia F, et al. Molecular findings among patients referred for clinical whole-exome sequencing. JAMA. 2014 Nov 12;312(18):1870-9. http://dx.doi.org/10.1001/jama.2014.14601.

70. Pereira RR, Pinto IP, Minasi LB, et al. Screening for intellectual disability using high-resolution CMA technology in a retrospective cohort from central Brazil. PLoS ONE. 2014 Jul 25;9(7):e103117. http://dx.doi.org/10.1371/journal.pone.0103117. PMID: 25061755.

71. Tao VQ, Chan KYK, Chu YWY, et al. The clinical impact of chromosomal microarray on paediatric care in Hong Kong. PLoS ONE. 2014 Oct 15;9(10). http://dx.doi.org/10.1371/journal.pone.0109629.

72. Soden SE, Saunders CJ, Willig LK, et al. Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Sci Transl Med. 2014 Dec 3;6(265):265ra168. http://dx.doi.org/10.1126/scitranslmed.3010076. PMID: 25473036.

73. Bartnik M, Nowakowska B, Derwinska K, et al. Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. J Appl Genet. 2014 Feb;55(1):125-44.

74. Boggula VR, Shukla A, Danda S, et al. Clinical utility of multiplex ligation-dependent probe amplification technique in identification of aetiology of unexplained mental retardation: A study in 203 indian patients. Ind J Med Res. 2014 Jan;66-75.

75. Byeon JH, Shin E, Kim GH, et al. Application of array-based comparative genomic hybridization to pediatric neurologic diseases. Yonsei Med J. 2014 Jan;55(1):30-6.

76. Chaudhary AG, Hussein IR, Abuzenadah A, et al. Molecular diagnosis of fragile X syndrome using methylation sensitive techniques in a cohort of patients with intellectual disability. Pediatr Neurol. 2014 Apr;50(4):368-76.

77. Cheon CK, Sohn YB, Ko JM, et al. Identification of KMT2D and KDM6A mutations by exome sequencing in Korean patients with Kabuki syndrome. J Hum Genet. 2014 Jun;59(6):321-5.

78. Chong WW, Lo IF, Lam ST, et al. Performance of chromosomal microarray for patients with intellectual disabilities/developmental delay, autism, and multiple congenital anomalies in a Chinese cohort. Mol Cytogenet. 2014 May 23;7(1):34.

79. Coutton C, Dieterich K, Satre V, et al. Array-CGH in children with mild intellectual disability: a population-based study. Eur J Pediatr. 2014 Jul 3;Epub ahead of print. PMID: 24985125.

80. Dubourg C, Bonnet-Brilhault F, Toutain A, et al. Identification of nine new rai1-truncating mutations in smith-magenis syndrome patients without 17p11.2 deletions. Mol Syndromol. 2014 Feb;5(2):57-64.

81. Fatima T, Zaidi SA, Sarfraz N, et al. Frequency of FMR1 gene mutation and CGG repeat polymorphism in intellectually disabled children in Pakistan. Am J Med Genet A. 2014 May;164(5):1151-61.

82. Gilissen C, Hehir-Kwa JY, Thung DT, et al. Genome sequencing identifies major causes of severe intellectual disability. Nature. 2014;511(7509):344-7.

83. Helsmoortel C, Vandeweyer G, Ordoukhanian P, et al. Challenges and opportunities in the investigation of unexplained intellectual disability using family based whole exome sequencing. Clin Genet. 2014 Aug 1;Epub ahead of print. PMID: 25081361.

84. Huguet G, Nava C, Lemiere N, et al. Heterogeneous pattern of selective pressure for PRRT2 in human populations, but no association with autism spectrum disorders. PLoS ONE. 2014;9(3):e88600. PMID: 24594579.

85. Lee JS, Byun CK, Kim H, et al. Clinical and mutational spectrum in Korean patients with Rubinstein-Taybi syndrome: The spectrum of brain MRI abnormalities. Brain Dev. 2014 Aug 6;Epub ahead of print. PMID: 25108505.

86. Medina A, Pineros L, Arteaga C, et al. Multiplex ligation-dependent probe amplification to subtelomeric rearrangements in idiopathic intellectual disability in Colombia. Pediatr Neurol. 2014 Mar;50(3):250-4.

87. Nicholl J, Waters W, Mulley JC, et al. Cognitive deficit and autism spectrum disorders: prospective diagnosis by array CGH. Pathology (Phila). 2014 Jan;46(1):41-5. PMID: 24300712.

88. Palmer E, Speirs H, Taylor PJ, et al. Changing interpretation of chromosomal microarray over time in a community cohort with intellectual disability. Am J Med Genet A. 2014 Feb;164A(2):377-85. PMID: 24311194.

89. Stobbe G, Liu Y, Wu R, et al. Diagnostic yield of array comparative genomic hybridization in adults with autism spectrum disorders. Genet Med. 2014 Jan;16(1):70-7.

90. Tucker T, Zahir FR, Griffith M, et al. Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. Eur J Hum Genet. 2014 Jun;22(6):792-800.

91. Tuysuz B, Kartal N, Erener-Ercan T, et al. Prevalence of prader-willi syndrome among infants with hypotonia. J Pediatr. 2014;164(5):1064-7. PMID: 24582009.

92. Utine GE, Haliloglu G, Volkan-Salanci B, et al. Etiological yield of SNP microarrays in idiopathic intellectual disability. Eur J Paediatr Neurol. 2014 May;18(3):327-37. Epub 2014 Jan 25. PMID: 24508361.

93. Uwineza A, Caberg JH, Hitayezu J, et al. Array-CGH analysis in Rwandan patients presenting development delay/intellectual disability with multiple congenital anomalies. BMC Med Genet. 2014;15(1):79. PMID: 25016475.

94. Vergult S, Van Binsbergen E, Sante T, et al. Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations. Eur J Hum Genet. 2014 May;22(5):652-9. PMID: 24105367.

95. Willemsen MH, Kleefstra T. Making headway with genetic diagnostics of intellectual disabilities. Clin Genet. 2014 Feb;85(2):101-10. PMID: 23895455.

96. Wiszniewska J, Bi W, Shaw C, et al. Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. Eur J Hum Genet. 2014 Jan;22(1):79-87. PMID: 23695279.

97. Ahn JW, Bint S, Bergbaum A, et al. Array CGH as a first line diagnostic test in place of karyotyping for postnatal referrals - results from four years' clinical application for over 8,700 patients. Mol Cytogenet. 2013;6(1):16.

98. Battaglia A, Doccini V, Bernardini L, et al. Confirmation of chromosomal microarray as a first-tier clinical diagnostic test for individuals with developmental delay, intellectual disability, autism spectrum disorders and dysmorphic features. Eur J Paediatr Neurol. 2013 Nov;17(6):589-99.

99. Behjati F, Ghasemi Firouzabadi S, Sajedi F, et al. Identification of chromosome abnormalities in subtelomeric regions using Multiplex Ligation Dependent Probe Amplification (MLPA) Technique in 100 Iranian patients with idiopathic mental retardation. Iran Red Crescent Med J. 2013 Oct;15(10):e8221.

100. Behjati F, Firouzabadi SG, Kariminejad R, et al. Genomic characterization of some Iranian children with idiopathic mental retardation using array comparative genomic hybridization. Indian J Hum Genet. 2013 Oct-Dec;19(4):443-8.

101. Del Carmen Montes C, Sturich A, Chaves A, et al. Clinical fndings in 32 patients with 22qll.2 microdeletion attended in the city of Cordoba, Argentina. Arch Argent Pediatr. 2013 Oct;111(5):423-7. PMID: 24092030.

102. Doherty E, O'Connor R, Zhang A, et al. Developmental delay referrals and the roles of Fragile X testing and molecular karyotyping: a New Zealand perspective. Mol Med Rep. 2013 May;7(5):1710-4. PMID: 23525284.

103. Esposito G, Ruggiero R, Savarese G, et al. A 15-year case-mix experience for fragile X syndrome molecular diagnosis and comparison between conventional and alternative techniques leading to a novel diagnostic procedure. Clin Chim Acta. 2013 Feb 18;85-9. PMID: 23279920.

104. Essop FB, Krause A. Diagnostic, carrier and prenatal genetic testing for fragile X syndrome and other FMR-1-related disorders in Johannesburg, South Africa: A 20-year review. S Afr Med J. 2013;103:994-8. PMID: 24300646.

105. Fan YS, Ouyang X, Peng J, et al. Frequent detection of parental consanguinity in children with developmental disorders by a combined CGH and SNP microarray. Mol Cytogenet. 2013;6(1):38.

106. Halder A, Jain M, Chaudhary I, et al. Fluorescence In Situ Hybridization (FISH) using non-commercial probes in the diagnosis of clinically suspected microdeletion syndromes. Ind J Med Res. 2013 Aug;138:135-42.

107. Jain S, Chowdhury V, Juneja M, et al. Intellectual disability in Indian children: Experience with a stratified approach for etiological diagnosis. Indian Pediatr. 2013 Dec;50(12):1125-30.

108. Jorge P, Oliveira B, Marques I, et al. Development and validation of a multiplex-PCR assay for X-linked intellectual disability. BMC Med Genet. 2013;80. PMID: 23914978.

109. Kashevarova AA, Skryabin NA, Cheremnykh AD, et al. Clinical and genetic analysis of idiopathic intellectual disability based on array comparative genomic hybridization. Zhurnal Nevrologii i Psihiatrii imeni S.S. Korsakova. 2013;2013(9):70-74.

110. Lee CG, Park SJ, Yun JN, et al. Array-based comparative genomic hybridization in 190 Korean patients with developmental delay and/or intellectual disability: a single tertiary care university center study. Yonsei Med J. 2013;54(6):1463-70.

111. Marano RM, Mercurio L, Kanter R, et al. Risk assessment models in genetics clinic for array comparative genomic hybridization: clinical information can be used to predict the likelihood of an abnormal result in patients. J Pediat Genet. 2013;2(1):25-31.

112. Mundhofir FE, Nillesen WM, Van Bon BW, et al. Subtelomeric chromosomal rearrangements in a large cohort of unexplained intellectually disabled individuals in Indonesia: A clinical and molecular study. Indian J Hum Genet. 2013 Apr;19(2):171-8. PMID: 24019618.

113. Nicholl J, Waters W, Suwalski S, et al. Epilepsy with cognitive deficit and autism spectrum disorders: prospective diagnosis by array CGH. Am J Med Genet B Neuropsychiatr Genet. 2013 Jan;162B(1):24-35. Epub 2012 Nov 26. PMID: 23184456.

114. Pohovski LM, Dumic KK, Odak L, et al. Multiplex ligation-dependent probe amplification workflow for the detection of submicroscopic chromosomal abnormalities in patients with developmental delay/intellectual disability. Mol Cytogenet. 2013;6(1):7.

115. Pratt DW, Warner JV, Williams MG. Genotyping FOXG1 mutations in patients with clinical evidence of the FOXG1 syndrome. Mol Syndromol. 2013 Jan;3(6):284-7.

116. Qiao Y, Tyson C, Hrynchak M, et al. Clinical application of 2.7M Cytogenetics array for CNV detection in subjects with idiopathic autism and/or intellectual disability. Clin Genet. 2013 Feb;83(2):145-54. PMID: 22369279.

117. Rodriguez-Revenga L, Vallespin E, Madrigal I, et al. A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. Gene. 2013 May 25;521(1):82-6. PMID: 23524024.

118. Saad K, Abdelrahman AA, Abdallah AM, et al. Clinical and neuropsychiatric status in children with Williams-Beuren Syndrome in Upper Egypt. Asian J Psychiatry. 2013 Dec;6(6):560-5. PMID: 24309873.

119. Shoukier M, Klein N, Auber B, et al. Array CGH in patients with developmental delay or intellectual disability: are there phenotypic clues to pathogenic copy number variants? Clin Genet. 2013 Jan;83(1):53-65. PMID: 22283495.

120. Sorte HS, Gjevik E, Sponheim E, et al. Copy number variation findings among 50 children and adolescents with autism spectrum disorder. Psychiatr Genet. 2013 Apr;23(2):61-9. PMID: 23277134.

121. Tos T, Vurucu S, Karkucak M, et al. Subtelomeric fish findings in turkish patients with idiopathic mental retardation. Genet Couns. 2013;24(3):259-64. PMID: 24341139.

122. Vallespin E, Palomares Bralo M, Mori MA, et al. Customized high resolution CGH-array for clinical diagnosis reveals additional genomic imbalances in previous well-defined pathological samples. Am J Med Genet A. 2013 Aug;161A(8):1950-60. PMID: 23798500.

123. Vallespin E, Palomares Bralo M, Mori MA, et al. Customized high resolution CGH-array for clinical diagnosis reveals additional genomic imbalances in previous well-defined pathological samples. Am J Med Genet A. 2013 Aug;161(8):1950-60. PMID: 23798500.

124. Vorsanova SG, Iourov IY, Kurinnaya OS, et al. Genomic abnormalities in children with mental retardation and autism: the use of comparative genomic hybridization in situ (HRCGH) and molecular karyotyping with DNA-microchips (array CGH). Zhurnal Nevrologii i Psihiatrii imeni S.S. Korsakova. 2013;2013(8):46-9.

125. Winarni TI, Utari A, Mundhofir FE, et al. Fragile X syndrome: Clinical, cytogenetic and molecular screening among autism spectrum disorder children in indonesia. Clin Genet. 2013 Dec;84(6):577-80.

126. Zarate YA, Dwivedi A, Bartel FO, et al. Clinical utility of the X-chromosome array. Am J Med Genet A. 2013 Jan;161A(1):120-30. PMID: 23208842.

127. Aradhya S, Lewis R, Bonaga T, et al. Exon-level array CGH in a large clinical cohort demonstrates increased sensitivity of diagnostic testing for Mendelian disorders. Genet Med. 2012 Jun;14(6):594-603. PMID: 22382802.

128. Dos Santos SR, Freire-Maia DV. Absence of subtelomeric rearrangements in selected patients with mental retardation as assessed by multiprobe T FISH. J Negat Results Biomed. 2012;11:16. PMID: 23259705.

129. Hochstenbach R, Poot M, Nijman IJ, et al. Discovery of variants unmasked by hemizygous deletions. Eur J Hum Genet. 2012 Jul;20(7):748-53. PMID: 22258528.

130. Iourov IY, Vorsanova SG, Kurinnaia OS, et al. Molecular karyotyping by array CGH in a Russian cohort of children with intellectual disability, autism, epilepsy and congenital anomalies. Mol Cytogenet. 2012;5(1):46. PMID: 23272938.

131. McGrew SG, Peters BR, Crittendon JA, et al. Diagnostic yield of chromosomal microarray analysis in an autism primary care practice: which guidelines to implement? J Autism Dev Disord. 2012 Aug;42(8):1582-91. PMID: 22089167.

132. Rafati M, Seyyedaboutorabi E, Ghadirzadeh MR, et al. "Familial" versus "Sporadic" intellectual disability: contribution of common microdeletion and microduplication syndromes. Mol Cytogenet. 2012;5(1):9. PMID: 22283845.

133. Rafati M, Ghadirzadeh MR, Heshmati Y, et al. "Familial" versus "sporadic" intellectual disability: contribution of subtelomeric rearrangements. Mol Cytogenet. 2012;5(1):4. PMID: 22260313.

134. Splendore A, da Rocha KM, Takahashi VN, et al. Centro de estudos do genoma humano: Our six-year experience on the molecular diagnosis of rett syndrome. Revista Neurociencias. 2012;20(2):194-9.

135. Tos T, Karaman A, Aksoy A, et al. Structural chromosomal abnormalities in patients with mental retardation and/or multiple congenital anomalies: A new series of 24 patients. Genet Couns. 2012;23(2):289-96. PMID: 22876589.

136. Tzetis M, Kitsiou-Tzeli S, Frysira H, et al. The clinical utility of molecular karyotyping using high-resolution array-comparative genomic hybridization. Expert Rev Mol Diagn. 2012 Jun;12(5):449-57. PMID: 22702362.

137. Utine GE, Kiper PO, Alanay Y, et al. Searching for copy number changes in nonsyndromic X-linked intellectual disability. Mol Syndromol. 2012 Jan;2(2):64-71. PMID: 22511893.

138. Bremer A, Giacobini M, Eriksson M, et al. Copy number variation characteristics in subpopulations of patients with autism spectrum disorders. Am J Med Genet B Neuropsychiatr Genet. 2011 Mar;156(2):115-24. PMID: 21302340.

139. Bruno DL, White SM, Ganesamoorthy D, et al. Pathogenic aberrations revealed exclusively by single nucleotide polymorphism (SNP) genotyping data in 5000 samples tested by molecular karyotyping. J Med Genet. 2011 Dec;48(12):831-9. PMID: 22039585.

140. Hannibal MC, Buckingham KJ, Ng SB, et al. Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. Am J Med Genet A. 2011 Jul;155(7):1511-6. PMID: 21671394.

141. Hayashi S, Imoto I, Aizu Y, et al. Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. J Hum Genet. 2011 Feb;56(2):110-24. PMID: 20981036.

142. Rana KS, Holla RG. To determine the frequency of subtelomeric abnormalities in children with idiopathic mental retardation. Med J Armed Forces India. 2011 Oct;67(4):326-8.

143. Roesser J. Diagnostic yield of genetic testing in children diagnosed with autism spectrum disorders at a regional referral center. Clin Pediatr (Phila). 2011 Sep;50(9):834-43. Epub 2011 Apr 27. PMID: 21525079.

144. Rooms L, Vandeweyer G, Reyniers E, et al. Array-based MLPA to detect recurrent copy number variations in patients with idiopathic mental retardation. Am J Med Genet A. 2011 Feb;155(2):343-8. PMID: 21271651.

145. Shawky RM, El-Baz F, Elsobky ES, et al. Screening for subtle chromosomal rearrangements in an Egyptian sample of children with unexplained mental retardation. Egypt J Med Hum Genet. 2011 May;12(1):63-8.

146. Wincent J, Anderlid BM, Lagerberg M, et al. High-resolution molecular karyotyping in patients with developmental delay and/or multiple congenital anomalies in a clinical setting. Clin Genet. 2011 Feb;79(2):147-57. PMID: 20486943.

147. Dave U, Shetty D. A PCR-based screening method for rapid detection and genetic counseling in Fragile-X syndrome. J Prenatal Diagn Ther. 2010 Jan-Jun;1(1):26-30.

148. Ezugha H, Anderson CE, Marks HG, et al. Microarray analysis in children with developmental disorder or epilepsy. Pediatr Neurol. 2010 Dec;43(6):391-4. PMID: 21093728.

149. Gervasini C, Mottadelli F, Ciccone R, et al. High frequency of copy number imbalances in Rubinstein-Taybi patients negative to CREBBP mutational analysis. Eur J Hum Genet. 2010 Jul;18(7):768-75. PMID: 20125191.

150. Manolakos E, Vetro A, Kefalas K, et al. The use of array-CGH in a cohort of Greek children with developmental delay. Mol Cytogenet. 2010;3(1):Article no. 22. PMID: 21062444.

151. Muscarella LA, Guarnieri V, Sacco R, et al. Candidate gene study of HOXB1 in autism spectrum disorder. Mol Autism. 2010;1:9.

152. Schaefer GB, Starr L, Pickering D, et al. Array comparative genomic hybridization findings in a cohort referred for an autism evaluation. J Child Neurol. 2010 Dec;25(12):1498-503. PMID: 20729506.

153. Shen Y, Dies KA, Holm IA, et al. Clinical genetic testing for patients with autism spectrum disorders. Pediatrics. 2010 Apr;125(4):e727-35. PMID: 20231187.

154. Siggberg L, Ala-Mello S, Jaakkola E, et al. Array CGH in molecular diagnosis of mental retardation - a study of 150 Finnish patients. Am J Med Genet A. 2010 Jun;152A(6):1398-410. PMID: 20503314.

155. Xiang B, Zhu H, Shen Y, et al. Genome-wide oligonucleotide array comparative genomic hybridization for etiological diagnosis of mental retardation: a multicenter experience of 1499 clinical cases. J Mol Diagn. 2010 Mar;12(2):204-12. PMID: 20093387.

156. Auber B, Bruemmer V, Zoll B, et al. Identification of subtelomeric genomic imbalances and breakpoint mapping with quantitative PCR in 296 individuals with congenital defects and/or mental retardation. Mol Cytogenet. 2009;2(1):10.

157. Baris I, Battaloglu E. A multiplexed ARMS-PCR approach for the detection of common MECP2 mutations. Genet Test Mol Biomarkers. 2009 Feb;13(1):19-22. PMID: 19309269.

158. Bhowmik DA, Dutta S, Chatterjee A, et al. Screening for fragile X syndrome among neurobehavioural patients from Kolkata, Eastern India. J Clin Diagn Res. 2009;3(1):1266-73.

159. Bucan M, Abrahams BS, Wang K, et al. Genome-wide analyses of exonic copy number variants in a family-based study point to novel autism susceptibility genes. PLoS Genet. 2009 Jun;5(6):e1000536. PMID: 19557195.

160. Cho EH, Park BY, Cho JH, et al. Comparing two diagnostic laboratory tests for several microdeletions causing mental retardation syndromes: multiplex ligation-dependent amplification vs fluorescent in situ hybridization. Korean J Lab Med. 2009 Feb;29(1):71-6. PMID: 19262082.

161. Cusco I, Medrano A, Gener B, et al. Autism-specific copy number variants further implicate the phosphatidylinositol signaling pathway and the glutamatergic synapse in the etiology of the disorder. Hum Mol Genet. 2009;18(10):1795-804. PMID: 19246517.

162. Dutta S, Das M, Bhowmik AD, et al. Screening of rural children in west bengal for Fragile-X syndrome. Ind J Med Res. 2009 Dec;130(6):714-9. PMID: 20090132.

163. Friedman JM, Adam S, Arbour L, et al. Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics. 2009 Nov 16;10:526. PMID: 19917086.

164. Gijsbers AC, Lew JY, Bosch CA, et al. A new diagnostic workflow for patients with mental retardation and/or multiple congenital abnormalities: test arrays first. Eur J Hum Genet. 2009 Nov;17(11):1394-402. PMID: 19436329.

165. Giorda R, Bonaglia MC, Beri S, et al. Complex segmental duplications mediate a recurrent dup(X)(p11.22-p11.23) Associated with Mental Retardation, Speech Delay, and EEG Anomalies in Males and Females. Am J Hum Genet. 2009 Sep 11;85(3):394-400. PMID: 19716111.

166. Hochstenbach R, van Binsbergen E, Engelen J, et al. Array analysis and karyotyping: Workflow consequences based on a retrospective study of 36,325 patients with idiopathic developmental delay in the Netherlands. Eur J Med Genet. 2009 Jul-Aug;52(4):161-9. PMID: 19362174.

167. Koolen DA, Pfundt R, de Leeuw N, et al. Genomic microarrays in mental retardation: a practical workflow for diagnostic applications. Hum Mutat. 2009 Mar;30(3):283-92. PMID: 19085936.

168. McMullan DJ, Bonin M, Hehir-Kwa JY, et al. Molecular karyotyping of patients with unexplained mental retardation by SNP arrays: A multicenter study. Hum Mutat. 2009 Jul;30(7):1082-92. PMID: 19388127.

169. Shahdadpuri R, Lambert D, Lynch SA. Diagnostic outcome following routine genetics clinic referral for the assessment of global developmental delay. Ir Med J. 2009 May;102(5):146-8. PMID: 19623810.

170. Utine GE, Celik T, Alanay Y, et al. Subtelomeric rearrangements in mental retardation: Hacettepe University experience in 130 patients. Turk J Pediatr. 2009;51(3):199-206. PMID: 19817261.

171. Mandal K, Boggula VR, Borkar M, et al. Use of multiplex ligation-dependent probe amplification (MLPA) in screening of subtelomeric regions in children with idiopathic mental retardation. Indian J Pediatr. 2009 Oct;76(10):1027-31. PMID: 19907935.

172. Baris HN, Tan WH, Kimonis VE, et al. Diagnostic utility of array-based comparative genomic hybridization in a clinical setting. Am J Med Genet A. 2007 Nov 1;143A(21):2523-33. PMID: 17910064.

173. de Souza DH, Moretti-Ferreira D, de Souza Rugolo LM. Flourescent In Situ Hybridization (FISH) as a diagnostic tool for Williams-Beuren Syndrome. Genet Mol Biol. 2007;30(1):17-20.

174. Newman WG, Hamilton S, Ayres J, et al. Array comparative genomic hybridization for diagnosis of developmental delay: an exploratory cost-consequences analysis. Clin Genet. 2007 Mar;71(3):254-9. PMID: 17309648.

175. Sandrin-Garcia P, Richieri-Costa A, Tajara EH, et al. Flourescence in situ hybridization (FISH) screening for the 22q11.2 deletion in patients with clinical features of velocardiofacial syndrome but without cardiac anomalies. Genet Mol Biol. 2007;30(1):21-4.

176. van Hagen JM, Eussen HJ, van Schooten R, et al. Comparing two diagnostic laboratory tests for Williams syndrome: fluorescent in situ hybridization versus multiplex ligation-dependent probe amplification. Genet Test. 2007;11(3):321-7. PMID: 17949295.

177. Coupry I, Monnet L, Attia AA, et al. Analysis of CBP (CREBBP) gene deletions in Rubinstein-Taybi syndrome patients using real-time quantitative PCR. Hum Mutat. 2004 Mar;23(3):278-84. PMID: 14974086.

178. Kleefstra T, Yntema HG, Nillesen WM, et al. MECP2 analysis in mentally retarded patients: implications for routine DNA diagnostics. Eur J Hum Genet. 2004 Jan;12(1):24-8. PMID: 14560307.

179. Vermeesch JR, Fiegler H, de Leeuw N, et al. Guidelines for molecular karyotyping in constitutional genetic diagnosis. Eur J Hum Genet. 2007 Nov;15(11):1105-14. PMID: 17637806.

180. Finsterer J, Harbo HF, Baets J, et al. EFNS guidelines on the molecular diagnosis of mitochondrial disorders. Eur J Neurol. 2009 Dec;16(12):1255-64. PMID: 19950421.

181. National Institute for Health and Clinical Excellence (NICE). Autism. Recognition, referral and diagnosis of children and young people on the autism spectrum. London (UK): National Institute for Health and Clinical Excellence (NICE); 2011 Sep. 51 p. (Clinical guideline; no.128).

182. Singapore Ministry of Health. Autism spectrum disorders in pre-school children. Singapore: Singapore Ministry of Health; 2010 Mar. 140 p.

183. Kearney HM, South ST, Wolff DJ, et al. American College of Medical Genetics recommendations for the design and performance expectations for clinical genomic copy number microarrays intended for use in the postnatal setting for detection of constitutional abnormalities. Genet Med. 2011 Jul;13(7):676-9. PMID: 21681105.

184. Ramsden SC, Clayton-Smith J, Birch R, et al. Practice guidelines for the molecular analysis of Prader-Willi and Angelman syndromes. BMC Med Genet. 2010;70. PMID: 20459762.

185. McConkieRosell A, Abrams L, Finucane B, et al. Recommendations from multi-disciplinary focus groups on cascade testing and genetic counseling for fragile X-associated disorders. J Genet Couns. 2007 Oct;16(5):593-606. PMID: 17497108.

186. South ST, Lee C, Lamb AN, et al. ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: Revision 2013. Genet Med. 2013 Nov;15(11):901-9. http://dx.doi.org/10.1038/gim.2013.129. PMID: 24071793.

187. Vermeesch JR, Brady PD, Sanlaville D, et al. Genome-wide arrays: quality criteria and platforms to be used in routine diagnostics. Hum Mutat. 2012 Jun;33(6):906-15. http://dx.doi.org/10.1002/humu.22076. PMID: 22415865.

188. Hanemaaijer NM, Sikkema-Raddatz B, van der Vries G,et al. Practical guidelines for interpreting copy number gains detected by high-resolution array in routine diagnostics. Eur J Hum Genet. 2012 Feb;20(2):161-5. PMID: 21934709.

189. Neul JL, Kaufmann WE, Glaze DG, et al. Rett syndrome: Revised diagnostic criteria and nomenclature. Ann Neurol. 2010 Dec;68(6):944-50. http://dx.doi.org/10.1002/ana.22124. PMID: 21154482.

190. Finucane B, Abrams L, Cronister A, et al. Genetic counseling and testing for FMR1 gene mutations: practice guidelines of the National Society of Genetic Counselors. J Genet Couns. 2012 Dec;21(6):752-60. PMID: 22797890.

191. Hochstenbach R, Buizer-Voskamp JE, Vorstman JA, et al. Genome arrays for the detection of copy number variations in idiopathic mental retardation, idiopathic generalized epilepsy and neuropsychiatric disorders: lessons for diagnostic workflow and research. Cytogenet Genome Res. 2011;135(3-4):174-202. PMID: 22056632.

192. Sagoo GS, Butterworth AS, Sanderson S, et al. Array CGH in patients with learning disability (mental retardation) and congenital anomalies: Updated systematic review and meta-analysis of 19 studies and 13,926 subjects. Genet Med. 2009 Mar;11(3):139-46. PMID: 19367186.

193. New NIH-funded resource focuses on use of genomic variants in medical care. Bethesda (MD): National Institutes of Health (NIH); 2013 Sep 25. http://www.nih.gov/news/health/sep2013/nhgri-25.htm. Accessed 2014 Dec 04.

194. Agency for Healthcare Research and Quality (AHRQ). Methods guide for medical test reviews [AHRQ Publication No. 12-EHC017]. Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2012 Jun. 188 p. http://effectivehealthcare.ahrq.gov/ehc/products/246/558/Methods-Guide-for-Medical-Test-Reviews_Full-Guide_20120530.pdf.

195. Leeflang MM, Deeks JJ, Gatsonis C, et al. Systematic reviews of diagnostic test accuracy. Ann Intern Med. 2008 Dec 16;149(12):889-97. PMID: 19075208.

Appendix A. Literature Search Methods

Resources Searched

ECRI Institute information specialists searched the following databases for relevant information. Search terms and strategies for each resource appear below.

Name	Date Limits	Platform/Provider
Bibliographic Databases		
The Cochrane Central Register of Controlled Trials (CENTRAL)	2009 through August 27, 2014	Wiley
The Cochrane Database of Methodology Reviews (Methodology Reviews)	2009 through August 27, 2014	Wiley
The Cochrane Database of Systematic Reviews (Cochrane Reviews)	2009 through August 27, 2014	Wiley
Database of Abstracts of Reviews of Effects (DARE)	2009 through August 27, 2014	Wiley
EMBASE (Excerpta Medica)	2009 through August 12, 2014	Embase.com
Health Technology Assessment Database (HTA)	2009 through August 27, 2014	Wiley
MEDLINE	2009 through August 12, 2014	Embase.com
PUBMED (PreMEDLINE)	Searched August 13, 2014	NLM
U.K. National Health Service Economic Evaluation Database (NHS EED)	2009 through August 27, 2014	Wiley
Gray Literature Resources		
American College of Medical Genetics and Genomics		ACMG
American Society of Human Genetics		ASHG
ClinicalTrials.gov	Searched August 26, 2014	NLM
EuroGentest – Clinical Utility Gene Cards	Searched August 28, 2014	Center for Human Genetics – University of Leuven
Genetic Test Registry	Searched throughout project	NCBI
Internet	Searched August 28, 2014	Google
MedGen	Searched March 7, 2014	NCBI
National Guideline Clearinghouse™ (NGC)	Searched August 27, 2014	AHRQ

Hand Searches of Journal and Gray Literature

Journals and supplements maintained in ECRI Institute's collections were reviewed. Nonjournal publications and conference proceedings from professional organizations, private agencies, and government agencies were also screened. Other mechanisms used to retrieve additional relevant information included review of bibliographies/reference lists from peer-reviewed and gray literature. (Gray literature consists of reports, studies, articles, and monographs produced by federal and local government agencies, private organizations,

educational facilities, consulting firms, and corporations. These documents do not appear in the peer-reviewed journal literature.)

Topic-Specific Search Terms

The search strategies employed combinations of free-text keywords as well as controlled vocabulary terms including (but not limited to) the following concepts. Strategies for each bibliographic database follow this table.

Medical subject headings (MeSH), EMTREE, and keywords

Concept	Controlled Vocabulary	Keywords
Conditions	**MEDLINE (MeSH)** 'Child development disorders, pervasive'/exp 'Chromosome disorders'/exp 'Developmental disabilities'/exp 'Intellectual disabilities'/exp **EMBASE (EMTREE)** 'autism'/exp 'cognitive defect':de 'congenital disorder':lnk 'intellectual impairment':de 'mental deficiency'/exp 'mental retardation malformation syndrome'/exp 'postnatal development'/exp	Autism Autistic ASD Asperger* PDD near/1 NOS PDDNOS Pervasive developmental disorder* Rett syndrome Developmental delay Developmental disability Intellectual delay Intellectual disability Mental retardation Neurocognitive impairment 1p36 deletion syndrome 9q deletion syndrome 17q21.31 deletion syndrome 18p minus syndrome 18q minus syndrome Alagille syndrome Angelman syndrome Charcot-Marie-Toothe syndrome Conotruncal anomaly face syndrome Cri du Chat syndrome DiGeorge syndrome Down syndrome Fragile x Fragile-x Happy puppet syndrome Jacobsen syndrome Kleefstra syndrome Koolen-de-vries syndrome Langer Gideon syndrome Miller-Dieker syndrome Prader-Willi PTEN deletion

Concept	Controlled Vocabulary	Keywords
		Rubinstein-Taybi Shrprintzen syndrome Smith-Magenis SRY deletion Velocardiofacial syndrome Wagr syndrome Williams syndrome Williams-Beuren syndrome Wolf-Hirschorn syndrome
Genetic Testing	**MEDLINE (MeSH)** 'chromosome disorders'/exp 'genetic techniques'/exp 'genetic testing'/exp 'microarray analysis'/exp 'oligonucleotide array sequence analysis':de 'comparative genomic hybridization':de 'molecular sequence data' 'sequence analysis, DNA':de 'sequence deletion'/genetics **EMBASE (EMTREE)** 'chromosome aberration'/exp – *note- this is a large category that encompasses the entire scope of this report.* 'epigenetics':de 'exome':de 'gene mutation'/exp 'gene sequencing':de 'genetic screening':de 'genetic procedures'/exp 'genome':de 'genome imprinting':de 'microarray analysis':de 'molecular diagnosis':de 'nucleic acid analysis'/exp	aCGH Array CGH Array genomic hybridization cDNA array cDNA microarray Chromosomal microarray analysis Chromosome deletion Chromosome duplication Comparative genomic hybridization Copy number Epigenetic* Gene chip* Genetic test* Imprinting Methylation Molecular diagnosis Next generation sequencing Nexgen NGS Single nucleotide polymorphism array SNP Whole exome Whole genome Affymetrix Agilent Technologies CytoScan Nimblegen Illumina
Specific genes and deletions		Copy number variants CNV CDKL5 CREBBP EP300 FMR1 FMR-1

Concept	Controlled Vocabulary	Keywords
		FOXG1 MECP2 TSC1 TSC2 UBE3A 10p deletion 20p deletion Deletion 1p36 Deletion 7q11.23 Deletion 10q13-q14 Deletion 17p11.2 Deletion 22q11.2 GJB6 deletion IDIC15 duplication Maternal deletion 15q11-q13 Paternal deletion 15q11-q13 STRC deletion

Search Strategies

EMBASE/MEDLINE (presented in Embase.com syntax)

Set Number	Concept	Search Statement
1	Genetic testing	'Chromosome aberration'/exp or (chromosom* NEAR/2 (duplicat* or deletion or 'copy number' or insertion))
2		'microarray analysis':de or 'nucleic acid analysis'/exp or 'molecular diagnosis':de or 'genetic screening':de or 'genetic procedures'/exp or 'array cgh' or 'aCGH' or 'CMA' or 'comparative genomic hybridization' or 'array genomic hybridization' or microarray or (molecular NEAR/2 diagnos*) or snp or 'single nucleotide polymorphism array' or (genetic NEAR/2 test*)
3		(exome:de OR genome:de) and 'gene sequencing':de
4		('whole exome' or 'whole genome') NEAR/3 sequencing
5		'next generation sequencing' or 'NGS'
6		'gene expression assay'/exp or 'gene chips' or 'cDNA array' or 'cDNA microarray' or 'genome imprinting':de or imprinting
7		Methylation or 'epigenetics':de or epigenetic*
8		#1 OR #2 OR #3 OR #4 OR #5 OR #6 OR #7
9	Conditions	Development* NEAR/2 (delay* or disabilit*)
10		'mental deficiency'/exp or (mental* NEAR/2 retard*) or (intellect* NEAR/2 (disabilit* or delay*))
11		(Neurocognitive NEAR/2 impair*) or 'cognitive defect':de or 'intellectual impairment':de
12		'Fragile X' or 'fragile-x' or 'mental retardation malformation syndrome'/exp
13		'autism'/exp or autistic* or autism or Asperger*:ti,ab or 'asd':ti,ab or 'rett syndrome' or 'pervasive developmental disorder' or 'PDD'
14	Specific syndromes (original)	'angelman syndrome'/exp OR 'happy puppet' OR 'prader-willi'/exp OR 'rubinstein-taybi'/exp OR 'smith magenis'/exp OR 'velocardiofacial syndrome'/exp OR 'digeorge syndrome'/exp OR 'shrprintzen syndrome' OR 'conotruncal anomaly face syndrome' OR 'williams syndrome'/exp OR 'williams-beuren syndrome'/exp
15	Specific syndromes – KI suggested	'kleefstra syndrome' OR 'miller-dieker syndrome' OR 'koolen-de vries syndomre' OR 'wagr syndrome' OR 'langer gideon syndrome' OR 'cri du chat syndrome' OR 'wolf-hirschorn syndrome' OR 'jacobsen syndrome' OR 'alagille syndrome' OR '1p36 deletion syndrome' OR '9q deletion syndrome' OR '17q21.31 deletion syndrome' OR '18p minus syndrome' OR '18q minus syndrome' OR 'sry deletion' OR 'pten deletion' OR 'charcot-marie-toothe syndrome'
16	Specific genes	ube3a OR fmr1 OR mecp2 OR cdkl5 OR foxg1 OR crebbp OR ep300
17	Combine sets	#9 OR #10 OR #11 OR #12 OR #13 OR #14 OR #15 OR #16
18	Removing rodents	#17 NOT (mouse*:ti OR mice:ti OR murine:ti OR rat:ti OR rodent:ti)
19	Diagnosis	'diagnostic test accuracy':de OR 'diagnosis':lnk OR 'receiver operating characteristic':de OR 'roc curve'/exp OR 'roc curve' OR 'sensitivity and specificity':de OR 'sensitivity' OR 'specficity' OR 'accuracy':de OR 'precision'/exp OR precision OR 'prediction and forecasting'/exp OR 'prediction and forecasting' OR 'diagnostic error'/exp OR 'diagnostic error' OR 'maximum likelihood method':de OR 'likelihood' OR 'predictive value'/exp OR 'predictive value' OR ppv OR (false OR true) NEAR/1 (positive OR negative)
20	Combine sets	#18 AND #19
21	Limit by keywords	#18 AND (idiopathic or (clinical NEAR/2 (valid* or util* or relevanc*)))
22	Combine sets	#20 OR #21
23	Limits	#22 NOT (prenatal:ti or maternal:ti)
24	Limit by publication and study type	#23 AND ('clinical article'/de OR 'clinical trial'/de OR 'cohort analysis'/de OR 'comparative study'/de OR 'controlled study'/de OR 'diagnostic test accuracy study'/de OR 'intermethod comparison'/de OR 'major clinical study'/de OR 'medical record review'/de OR 'practice guideline'/de OR 'prospective study'/de OR 'retrospective study'/de OR 'validation study'/de) AND ('Article'/it OR 'Article in Press'/it OR 'Conference Abstract'/it OR 'Conference Paper'/it OR 'Review'/it)

EMBASE.com Syntax:

*	=	truncation character (wildcard)
NEAR/*n*	=	search terms within a specified number (*n*) of words from each other in any order
NEXT/*n*	=	search terms within a specified number (*n*) of words from each other in the order specified
/	=	search as a subject heading
exp	=	"explodes" controlled vocabulary term (e.g., expands search to all more specific related terms in the vocabulary's hierarchy)
mj	=	denotes a term that has been searched as a major subject heading
:de	=	search in the descriptors field
:lnk	=	floating subheading
:it,pt.	=	source item or publication type
:ti.	=	limit to title
:ti,ab.	=	limit to title and abstract fields

PUBMED (PreMEDLINE)

Strategy: limits = human, English language

Set Number	Concept	Search Statement
1	Genetic testing	Chromosom* AND (aberration* OR duplicat* OR deletion* OR "copy number repeat" OR insertion*)
2		Microarray* OR "nucleic acid analysis" OR "molecular diagnosis" OR "molecular testing" OR "genetic screening" OR "array CGH" OR "aCGH" OR "CMA" OR "comparative genomic hybridization" OR "array genomic hybridization" OR "single nucleotide polymorphism array"
3		"whole genome sequencing" OR "whole exome sequencing"
4		"next generation sequencing" OR "NGS"
5		"gene expression assay" OR "gene chips" OR "cDNA array" OR "cDNA microarray" OR "genome imprinting" OR imprinting
6		Methylation OR epigenetic*
7		#1 OR #2 OR #3 OR #4 OR #5 OR #6 OR #7
8	Conditions	"developmental delay" OR "developmental deficiency"
9		"mental deficiency" OR "mental retardation" OR "mentally retarded" OR "intellectual disability" OR "intellectual delay" OR "intellectual impairment"
10		"neurocognitive impairment" OR "cognitive defect"
11		"Fragile X" OR "fragile-x"
12		autistic* OR autism OR Asperger*[tiab] OR 'asd' OR "rett syndrome" OR "pervasive developmental disorder" OR PDD
13		"angelman syndrome" OR "happy puppet" OR "prader-willi" OR "rubinstein-taybi" OR "smith Magenis" OR "velocardiofacial syndrome" OR "digeorge syndrome" OR "shrprintzen syndrome" OR "conotruncal anomaly face syndrome" OR "williams syndrome" OR "williams-beuren syndrome"
14		"kleefstra syndrome" OR "miller-dieker syndrome" OR "koolen-de vries syndome" OR "wagr syndrome" OR "langer gideon syndrome" OR "cri du chat syndrome" OR "wolf-hirschorn syndrome" OR "jacobsen syndrome" OR "alagille syndrome" OR "1p36 deletion syndrome" OR "9q deletion syndrome" OR "17q21.31 deletion syndrome" OR "18p minus syndrome" OR "18q minus syndrome" OR "sry deletion" OR "pten deletion" OR "charcot-marie-toothe syndrome"
		ube3a OR fmr1 OR mecp2 OR cdkl5 OR foxg1 OR crebbp OR ep300
16	Combine sets	#8 OR #9 OR #10 OR #11 OR #12 OR #13 OR #14 OR #15 #7 AND #16
17	Limit to in process	#16 AND (pubmednotmedline[sb] OR inprocess[sb] OR [publisher[sb])
18	Diagnosis	#17 AND (diagnos* OR detect* OR identif* OR "clinical validity" OR "clinical utility" OR "clinical relevancy")
19		#17 AND idiopathic
20		#18 OR #19
21		#20 AND English[la]
22		#21 NOT (prenatal OR fetal OR maternal)
23		#22 NOT (cancer* OR neoplasm* OR tumor* OR tumour* OR carcinoma* OR sarcoma* OR glioma*)
24		#23 AND (rat OR rats OR rodent* OR mouse OR mice OR murine OR hamster*)
25		#23 NOT #24

PubMed Syntax:

*	=	truncation character (wildcard)
[ti]	=	limit to title field
[tiab]	=	limit to title and abstract fields
[tw]	=	text word

Cochrane Library databases

Search Strategy	Search Statement	Number Identified
#1	MeSH descriptor: [Chromosome Disorders] explode all trees	645
#2	chromosom* near/2 (duplicat* or deletion or 'copy number' or insertion)	80
#3	MeSH descriptor: [Microarray Analysis] explode all trees	269
#4	MeSH descriptor: [Genetic Techniques] explode all trees	4,696
#5	MeSH descriptor: [Genetic Testing] explode all trees	494
#6	'array cgh' or 'aCGH' or 'CMA' or 'comparative genomic hybridization' or 'array genomic hybridization' or microarray or (molecular near/2 diagnos*) or snp or 'single nucleotide'	2,543
#7	('whole exome' or 'whole genome') near/3 sequencing	13
#8	'next generation sequencing' or 'NGS'	2,158
#9	'gene chips' or 'cDNA array' or 'cDNA microarray'	72
#10	Methylation or epigenetic*	358
#11	#1 or #2 or #3 or #4 or #5 or #6 or #7 or #8 or #9 or #10	9,411
#12	Development* near/2 (delay* or disabilit*)	1,404
#13	(mental* near/2 retard*) or (intellect* near/2 (disabilit* or delay*))	1,643
#14	MeSH descriptor: [Developmental Disabilities] explode all trees	453
#15	MeSH descriptor: [Intellectual Disability] explode all trees	1,020
#16	(Neurocognitive near/2 impair*)	83
#17	"happy puppet" or "conotruncal anomaly face syndrome" or "kleefstra syndrome" or "miller-dieker syndrome" or "koolen-de vries syndrome" or "WAGR syndrome" or "langer Gideon syndrome" or "cri du chat syndrome" or "alagille syndrome" or "1p36 deletion syndrome" or "9g deletion syndrome" or "17q21.31 deletion syndrome" or "18p minus syndrome" or "18q minus syndrome" or "sry deletion" or "pten deletion" or "characote-marie-toothe syndrome"	10
#18	ube3a or FMR1 or mecp2 or cdkl5 or foxg1 or crebbp or ep300	25
#19	"fragile x" or "fragile-x"	73
#20	MeSH descriptor: [Autistic Disorder] explode all trees	519
#21	MeSH descriptor: [Asperger Syndrome] explode all trees	39
#22	Autism or autistic	1,154
#23	#12 or #13 or #14 or #15 or #16 or #17 or #18 or #19 or #20 or #21 or #22	4,149
#24	#11 and #23 Publication Year from 2009 to 2014	306
#25	#24 and (diagnos* or "receiver operating characteristic" or "ROC" or sensitivity or specificity or accuracy or precision or prediction or predictive)	227
#26	#24 and ((true or false) near/1 (positive or negative))	31
#27	#24 and idiopathic	12
#28	#24 and (clinical near/2 (valid* or util* or relevanc*))	19
#29	#25 or #26 or #27 or #28	228

Cochrane Library Syntax:

* = truncation character (wildcard)

The Cochrane Library via the Wiley platform is menu-driven.

Appendix B. Genetic Testing Overview

DNA, Genes, and Development

DNA contains the information for constructing protein molecules that catalyze the cascade of biochemical reactions that govern the body's growth and development as well as serving as some of the basic building blocks of cellular structure. The DNA in our cells is arranged as two helical strands of linearly-arranged chains of building blocks called nucleotides. There are four types of nucleotides in DNA: adenine (A), thymine (T), guanine (G), and cytosine (C). Due to the inherent chemical properties of these individual nucleotides, adenine and thymine have high affinities and attract one another (bond) when in proximity, as do cytosine and guanine. In a complete, double-stranded DNA molecule, each cytosine on one strand is aligned with a guanine on the other, and each adenine nucleotide on one strand is paired with a thymine on the other. As such, the two strands interlock, or hybridize, with each other. Further chemical interactions between individual nucleotides cause this entire assembly to adopt its unique helical shape.[1,2]

DNA is contained in chromosomes, complex structures located within the nucleus (central area) of cells. Chromosomes come in pairs, one from each parent. Humans have 23 pairs of chromosomes (46 chromosomes total) distinguished by their unique size and shape under a microscope. They are numbered from 1 to 22, with an additional, 23rd, pair of sex chromosomes, denoted X and Y. Girls have two X chromosomes (denoted XX), whereas boys have one X and one Y (denoted XY). Genes are segments of the DNA molecule that contain the information for constructing a particular protein (sequence of amino acids), dictated by their unique sequence of nucleotides. An allele is a particular form, or version, of a gene. Each individual typically has two alleles for each gene, with some exceptions.[1,2]

As stated above, DNA contains the information for the construction of protein molecules, linear chains of basic biochemical molecules called amino acids. Protein molecules are a part of the basic building blocks of cellular structure, and they facilitate metabolic reactions for the maintenance, growth, and proliferation of cells. The carefully-coordinated growth and proliferation of different types of specialized cells (e.g., brain, muscle, kidney cells) and the processes allowing for their coordinated function are responsible for organism development and growth.[1] These processes are fundamentally dependent on activation of specific combinations of genes at the proper time, in much the same way that a well-played musical composition is dependent on the properly-timed activation of different musical notes.

Genetic Disorders

Genetic disorders are medical conditions resulting from abnormalities in DNA (genes) or the regulatory processes that govern gene activation. Changes in the number or structure of chromosomes or aberrations in the total amount of DNA may cause or increase the risk of congenital defects, developmental delays, behavioral disorders, and intellectual disabilities. For instance, Down syndrome, also known as trisomy 21, is a disorder causing mental retardation. It is caused by the presence of three copies of all or part of the 21st chromosome (an example of an aneuploidy, a condition marked by an abnormal number of chromosomes in each cell).[3] Other disorders, such as Rett syndrome, a developmental disorder associated with delayed development and intellectual disability, can result from a defect (or mutation) in a single gene.[4] In many cases, however, developmental disorders are caused by multiple genetic mutations. Autism, intellectual disabilities, and developmental delay, for instance, are often due to a combination of abnormalities in a number of different genes.[5] Typically, for these disorders, a given genetic

abnormality may confer an *increase in risk* for the disorder but do not by themselves cause the disorder. Possessing a greater number of risk-conferring genetic abnormalities will increase one's chances of having a disorder. Copy-number variations (CNVs) are another example of structural variation that can increase the risk of disease. CNVs are relatively large portions of the genome that have been deleted or duplicated on certain chromosomes, resulting in the cells having, respectively, fewer or more copies of certain segments of DNA. The role of CNVs in conferring risk for developmental disorders is a very active area of research.[6]

Understanding the tremendous heterogeneity in genetic makeup that can predispose individuals to disease is a major scientific hurdle that must be surmounted in order to develop more effective treatments, and potentially cures, for developmental disorders. Efforts to advance the field of genetic testing have focused on meeting this challenge.

Genetic Testing

Identifying the aberrations in genetic material that underlie disease states is a central task in better understanding disease etiology and for developing more effective treatments. Many different types of genetic tests exist that can reveal abnormalities in genetic information at varying levels of complexity, ranging from the gain or absence of entire chromosomes to mutations in a single nucleotide within a gene.

Cytogenetic testing (or chromosome testing) examines the number and structure of chromosomes. This type of testing can detect whether there are too many or too few of any one chromosome, or whether there is too much (gain) or too little (loss) of a particular portion of the chromosomes. Disorders such as Down syndrome (discussed above) and fragile X syndrome (mental retardation due to abnormalities of the X chromosome) are consequences of such abnormalities.

Karyotyping is a test involving the gross examination of chromosomes in a sample of cells. Karyotyping can reveal variations in the number of chromosomes (as in Down syndrome) and large-scale structural abnormalities in individual chromosomes. Chromosome banding methods are an extension of karyotyping. When treated with various dyes, different segments along the length of a chromosome stain to different degrees. This produces a banding pattern, like a series of stripes, perpendicular to the long axis of the chromosome. This banding pattern can be viewed under a microscope and provides a more detailed image of chromosome microstructure, making possible the identification of smaller-scale chromosomal abnormalities such as translocations (relocations of DNA segments from one chromosome to another), missing segments of DNA (deletions), and amplifications (multiple copies of chromosomal segments).[2,7]

An increasingly sophisticated panoply of tools from molecular biology has greatly enhanced the level of detail and rapidity with which genetic abnormalities can be detected and analyzed. These molecular-based genetic tests are the primary subjects of this report and we provide a brief survey of these below.

Fluorescence in situ hybridization (FISH) allows for visualization and mapping of specific genes or portions of genes. It can be used to identify where a particular gene falls within an individual's chromosomes. This is accomplished by preparing short sequences of single-stranded DNA (called probes) that are complimentary (in their nucleotide sequence) to the gene or portions of the gene of interest. These probes are then labeled by attaching one of a number of fluorescent dyes of a specific color. Because the probes are single-stranded, they can bind to complementary strands of DNA in the person's chromosomes, thus visually "tagging" the gene or gene segment of interest.[1,8]

Advanced Genetic Tests

In the past 10–15 years, genetic testing methods have greatly increased in sophistication. It is now possible not only to examine abnormalities in the number or structure of individual chromosomes, but also to examine the composition of large portions of the genome or even its entirety. Today, four major genome-wide assays are used to assess single nucleotide polymorphism (SNP) base variants and CNVs, ordered in terms of method sophistication and clinical utility:

- Array comparative genomic hybridization (aCGH)
- SNP microarray (array)
- Whole exome sequencing (WES)
- Whole genome sequencing (WGS)

Array CGH is a more recent genetic testing technology that allows the detection of very small chromosomal imbalances that cannot be identified through karyotyping (analysis of gross structure with use of a microscope). Very small changes in the amount of genetic information are referred to as microduplications (gain of genetic information) or microdeletions (loss of genetic information). Array CGH compares an individual's DNA with a control sample of DNA (typically from an individual not possessing the disorder of interest) and identifies differences between the two sets of DNA. Array CGH efficiently identifies deletions or duplications (imbalances) in an individual's DNA. This allows physicians and researchers to relate an individual's physical and mental characteristics (phenotype) to detailed profile of their genetic makeup (genotype).[9,10]

Because of the binding properties of DNA nucleotide base pairs, one strand of DNA will bind to (or "hybridize") with another strand having complimentary nucleotide sequence. This property forms the basis of the aCGH technique. A DNA microarray is comprised of thousands of short sequences of DNA, called "probes," arranged in a precise spatial grid on the surface of a glass slide, termed a DNA "chip." DNA from a patient is first "digested" with bacterial restriction enzymes that chop it up into short fragments (restriction enzymes are an evolutionary adaptation by bacteria to combat viral infection). These fragments are labeled with a fluorescent dye of a specific color. Reference DNA from another subject or multiple subjects with no known genetic abnormalities is also digested and labeled with fluorescent dye of another color (often, red and green dyes are used for the patient and control subjects, respectively). The two DNA samples, one from the patient and the other from the normal individual or group, are mixed together and applied to the surface of the chip. The applied fragments of DNA then hybridize with their complimentary probes, thus fluorescently labeling the spatial locations on the DNA chip containing the complimentary nucleotide sequences. The DNA chip and attached DNA test fragments can then be "read" by a microarray scanner which calculates the ratio of red and green fluorescence emitted from each location on the chip. Locations on the chip (representing specific segments of the DNA molecule) that have an equivalent degree of complimentary binding to the two patient samples will fluoresce yellow, indicating the patient has the same amount of genetic material as the control subjects in this segment of DNA. If the patient has a deletion at that segment, there will be more green fluorescence, while a duplication will result in more red fluorescence. Array CGH readings are quantified in terms of intensity data (of one color) alone (normalized at 0 for diploid), and so typically are used to determine duplications and deletions.[9-11]

Single nucleotide polymorphism microarrays (SNPs, or "snips") are genome positions where two distinct nucleotide residues exist on different alleles, with each one appearing in a

substantial portion of the human population. SNPs comprise a major portion of human genetic variability, with an estimate of 10 million SNPs in the human genome. SNP arrays are useful for identifying small variations between whole genomes. SNP arrays work on the same general principles as aCGH, but while DNA from both a patient and control subjects are used in aCGH, SNP arrays involve hybridization of only the patient's DNA. SNP arrays utilize two different oligonucleotides, one matching each of two variant alleles. The two alleles of a SNP are typically denoted A and B, and because each individual usually inherits one allele from each parent, the individual's genotype at a given SNP site will be AA, AB or BB. SNP arrays, therefore, can tell whether specific genetic variants are present and also provide the specific genotype of these variants with fluorescent intensity data (normalized at 0, 0.5 and 1 for AA, AB, and BB, respectively), throughout the entire genome.[12,13]

Originally, SNP arrays were designed for genome-wide association studies, being used to detect genotypes for thousands of single-nucleotide differences (polymorphisms) across an entire genome. More recent applications for SNP arrays include determining disease risk (similar to aCGH) and predicting or measuring the efficacy of drug therapies for individual patients. SNP arrays can also detect uniparental disomy (receiving two copies of a chromosome from one parent), consanguinity (offspring resulting from matings of close relatives), and copy number variation (CNVs).[12-14]

Whole-exome and whole-genome sequencing. DNA contains both coding regions (portions of the DNA that code for proteins, termed exons) and non-coding regions (termed introns). The entirety of the protein-coding regions of all DNA in a genome (all of the exons) is termed the "exome." The entirety of human DNA contains roughly 180,000 exons that comprise roughly 1% of the entire genome. These 180,000 exons are distributed throughout about 22,000 genes. Since exons code for proteins, they contain most of the errors that occur in DNA sequences that underlie genetic disorders.[15-17] Exome sequencing is therefore an efficient method of analyzing a patient's DNA to elucidate the underlying cause of many diseases and disabilities. WES uses genome-wide, targeted exon capture to define the precise sequence of individual nucleotides in the entire exome, called the "consensus sequence." This consensus sequence is then compared to standard and reference sequences representing what is normal (or genetically typical) of the overall population. Variations in an individual's DNA sequence relative to standards can then be identified to help assess the risk of disease or be used along with a patient's clinical assessment to either confirm or help establish a diagnosis. WES is of primary importance for identifying and analyzing base and copy variants, alike. Additionally, WES also examines DNA found within mitochondria, small structures within a cell that participate in providing usable energy for the body.[18-20]

WGS is a method for resolving the detailed nucleotide sequence of the entire genome. (Although the whole genome, or exome for WES, is theoretically sequenced, some regions are poorly sequenced or mapped to the genome, creating residual variability and imperfect continuous coverage.) WGS, therefore, has the ultimate potential to resolve almost the entirety of structural variation in a patient's genome, including intronic as well as exonic segments of DNA. Although introns do not code directly for proteins, they may affect the extraction of information from DNA in exonic regions and may have other regulatory functions on genetic networks.[21] Examining sequence variations in these regions has the potential to reveal additional contributors to disease. Even in otherwise healthy individuals, WES and WGS are expected to reveal genetic variations of potential clinical importance. Many clinicians have already acknowledged the

potential of WES and WGS to aid clinical diagnosis, identify the genetic underpinnings of rare diseases, and help inform studies in the biological basis of disease.[22-24]

Polymerase chain reaction (PCR) is a biochemical method for replicating strands of DNA. Beginning with a few strands of DNA, PCR can be used to generate thousands or millions of new copies in a relatively short time.[2,25] PCR makes use of DNA polymerases, a class of enzymes that operate within cells to create new DNA. DNA replication normally occurs when cells divide in a fashion that maintains the total complement of genetic material in each resulting cell constant. Once two strands of a DNA molecule are separated, a given molecule of DNA polymerase can attach to one of the strands and begin catalyzing the addition of complimentary nucleotides to that strand to once again form a complete segment of double-stranded DNA.[2,25] By exposing segments of DNA repeatedly to cycles of heating followed by cooling (thermal cycling) in the presence of DNA polymerase and short segments of complimentary DNA called primers, this process of DNA replication can be repeated many times over (usually 25–40 times), creating an exponential increase in the number of DNA strands. These thermal cycles normally consist of three stages: the first stage, carried out at about 95–98 °C, causes separation of the two strands of DNA. In the second stage, at a temperature of about 50–60 °C, the primers bind to the DNA template. Finally, the third stage, carried out at about 72 °C, facilitates the polymerization carried out by the DNA polymerase. PCR is indispensable in isolating and amplifying DNA segments for research purposes, as well as aiding in the diagnosis of genetic disorders, DNA fingerprinting, and detecting and identifying viruses or bacteria.[2,26]

Multiplex-PCR is a variation of the PCR technique that employs multiple primer sets within a single PCR mixture, producing different amplicons (a segment of replicated DNA or RNA) of varying sizes, each specific to a unique DNA sequence. Using PCR to target multiple genes at once provides additional information from a single test run that otherwise would require considerably more time and resources to perform.[27]

Quantitative PCR (qPCR) uses the PCR technique to amplify and quantify a specific DNA segment. Amplified DNA is detected during the PCR reaction in "real time." To detect products of qPCR reactions, fluorescent dyes that embed into double-stranded DNA or fluorescently labeled oligonucleotides (short, artificially-created segments of DNA) that activate upon binding a complementary sequence are commonly used. Quantitative PCR is carried out in a thermal cycler with the capacity to quantify the amount of fluorescence and thus the presence and amount of each type of DNA produced.[28,29]

Multiplex ligation-dependent probe amplification (MLPA) is a special instance of multiplex PCR. It can be used to detect small copy-number changes in DNA or RNA sequences, such as deletions or duplications within a single gene and is more sensitive than real-time PCR. Unlike multiplex PCR, MLPA allows for the detection of aberrant copy number in many genomic DNA sequences in a single PCR reaction. It can also detect methylation status of DNA segments. Methylation is a process by which certain genes can be inactivated within a cell (see discussion below). MLPA requires very little DNA and can discriminate sequences differing in only a single nucleotide. For laboratory purposes, it has the added advantage of being applicable to partially degraded DNA, as might be found in chemically-treated or preserved tissue samples.[30,31]

The MLPA process begins with an MLPA probe that contains two probe oligonucleotides, short segments of manufactured DNA complimentary to a sequence of interest in a sample of DNA. If the probe's target sequence is present in the sample DNA, the two probe oligonucleotides attach to the sample next to each other and are ligated (connected together). The

ligated probes (not the sample DNA) are then amplified in the multiplex PCR reaction. The relative amounts of amplification products generated by the probes then reflect the relative copy number (copy number variation) of target sequences.[30,31]

MLPA is a very fast and accurate way to detect genomic CNVs, deletions and insertions that are frequent contributors to disorders such as autism and Prader-Willi or Angelman syndromes. MLPA can easily discriminate the relative copy number of all exons within a gene simultaneously with high sensitivity.[30,31]

DNA methylation analysis is a biochemical process in which a methyl group is added to the cytosine or adenine DNA nucleotides causing the DNA sequence to be inaccessible to enzymes involved in transcribing information to form proteins. Methylation, therefore, essentially "locks" the gene in the off position. DNA methylation plays an important role in organism development and in the genesis of disease, acting as a major influence in epigenetic gene regulation (the activation and deactivation of subsets of the genome, oftentimes in response to environmental influences). Different diseases are associated with different methylation patterns across many genes. In developmental disorders, these patterns of gene regulation exert powerful influences over the developing organism. Identifying methylation patterns, or "signatures," throughout the genome can, in principle, provide a very powerful diagnostic tool. DNA methylation is detected by a variety of assays currently used in scientific research, including mass spectrometry, methylation-specific PCR, and whole genome bisulfite sequencing.[32,33]

Appendix C. Definition of Terms

The key terms used in this report are defined in this section. The definitions are from an Agency for Healthcare Research and Quality Evidence-based Practice Center method report we published, titled "Addressing Challenges in Genetic Test Evaluation. Evaluation Frameworks and Assessment of Analytic Validity."[34]

Analytic accuracy: the closeness of agreement between the result of a measurement and a true value of the measurand.

Analytic sensitivity: the ability of a test to effectively detect all true-positive specimens, as determined by a reference method. As it is more often used, this term is used for tests that yield a qualitative result.

Analytic specificity: the ability of a measurement procedure to measure solely the analyte of interest. Two important aspects of analytic specificity are interference by endogenous or exogenous substances other than the analyte of interest and cross-reactivity of the analytic system with substances other than the intended analyte of interest.

Analytic validity: how well a test performs in the laboratory—how well does the test measure the properties or characteristic it is intended to measure (e.g., a gene mutation)?

Assay linearity: the ability (within a given range) to provide results that are directly proportional to the concentration (amount) of the analyte in the test sample. Linearity of tests is established by testing a dilution series of a positive sample.

Clinical utility: the usefulness of a test and the value of its information to medical practice. If a test has utility, it means that the results of the test can be used to seek effective treatment or provide other concrete benefit.

Clinical validity (also known as diagnostic accuracy): the accuracy with which a test predicts the presence or absence of a clinical condition or predisposition.

Cross-reactivity: the reaction that an assay may have with analytes other than the ones it is designed to measure.

Diagnostic accuracy: see *clinical validity*.

Diagnostic sensitivity: the probability of a positive test result when disease is present.

Diagnostic specificity: the probability of a negative test result when disease is absent.

Gray literature: reports, studies, articles, and monographs produced by federal and local government agencies, private organizations, educational facilities, consulting firms, and corporations. These documents do not appear in the peer-reviewed journal literature.

Health outcomes: symptoms and conditions that patients can feel or experience, such as visual impairment, pain, dyspnea, impaired functional status or quality of life, and death.

Interference: may result from contamination, admixture, or presence of exogenous substances in samples, which can occur for a variety of reasons such as poor sampling; lack of sample stabilizer (if appropriate); cross-contamination during sample processing, inclusion of normal, non-diseased tissue with the diseased tissue of interest, tissue from a source additional to the desired sample (e.g., maternal cells obtained during fetal specimen collection), or failure to

remove exogenous substances (e.g., anticoagulants used during blood collection, residual reagents used during sample processing).

Intermediate outcomes: pathologic and physiologic measures that may precede or lead to health outcomes. For example, elevated blood cholesterol level is an intermediate outcome for coronary artery disease.

Precision: the closeness of agreement between independent results of measurements obtained under stipulated conditions. Precision is commonly determined by assessing repeatability and reproducibility (both of these terms are defined in this Appendix).

Recovery (as a term in the area of analytic validity): the measurable increase in analyte concentration or activity in a sample after adding a known amount of that analyte to the sample.

Reference range (also known as reference interval or normal values): the range of test values expected for a designated population of persons (e.g., 95 percent of people who are presumed to be healthy [or normal]).

Repeatability: replication of results when the assay is performed multiple times on a single specimen. Repeatability is also referred to as precision (in the term's narrow sense) when the test result is expressed quantitatively.

Reportable range of test results: the span of test-result values over which the laboratory can establish or verify the accuracy of the instrument or test system measurement response.

Reproducibility: the closeness of agreement between independent results of measurements obtained with the same assay method when as many known variables as possible (e.g., operators, instruments, reagent lots, day of the week, sites/laboratories) are tested for their effect on the assay result.

Robustness: the ability of a method to remain unaffected by small fluctuations in assay parameters; it is often assessed through inter-laboratory comparison studies or by varying parameters such as temperature and relative humidity to determine the operating range of the method.

Traceability: a property of the result of a measurement or the value of a standard that relates to stated references, usually national or international standards, through an unbroken chain of comparisons, all having stated uncertainties.

Uncertainty: a parameter associated with the result of a measurement that characterizes the dispersion of the values that could reasonably be attributed to the measurand; it is a formal quantitative statement of the confidence in the result of an assay.

Appendix D. Genetic Tests for Developmental Disabilities

Table D-1. Genetic tests for Angelman syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000303241.5	(AC) Atlanta Center: Medical Neurogenetics, LLC. Georgia 11D0703390 303241	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Skin|White blood cell prep	Decline to answer	Yes
GTR000320684.3	Ambry Genetics Ambry Genetics California 05D0981414 320684	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Dried blood spot (DBS) card|Isolated DNA|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000512791.2	Athena Diagnostics Inc Massachusetts 22D0069726 512791	ARHGEF9|ARX|CDKL5|CNTNAP2|FOXG1|GABRG2|GRIN2A|KCNT1|MECP2|NRXN1|PCDH19|PNKP|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYNGAP1|TCF4|TREX1|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000512796.2	Athena Diagnostics Inc Massachusetts 22D0069726 512796	ALDH7A1|ARFGEF2|ARHGEF9|ARX|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATRX|CACNA1A|CASK|CASR|CCDC88C|CDKL5|CHRNA2|CHRNA4|CHRNB2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COL18A1|COL4A1|CPT2|CSTB|CTSD|CUL4B|DCX|DEPDC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5|DNAJC5|EFHC1|EMX2|EPM2A|FGD1|FGFR3|FKRP|FKTN|FLNA|FOXG1|GABRA1|GABRB3|GABRD|GABRG2|GPC3|GPR56|GRIA3|GRIN2A|HSD17B10|KCNA1|KCNJ1|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|LAMA2|LARGE|LBR|LGI1|MBD5|ME2|MECP2|MEF2C|MFSD8|NHLRC1|NIPBL|NOTCH3|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PCDH19|PEX7|PHF6|PIGV|PLA2G6|PLP1|PNKP|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRRT2|RAB39B|RAB3GAP1|RAI1|RELN|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SERPINI1|SETBP1|SLC25A22|SLC2A1|SLC4A10|SLC9A6|SMC1A|SMC3|SMS|SNAP29|SPTAN1|SRPX2|STXBP1|SYNGAP1|SYP|TBC1D24|TBX1|TCF4|TPP1|TREX1|TSC1|TSC2|TUBA1A|TUBA8|TUBB2B|UBE3A|VPS13A|VPS13B|WDR62|ZEB2					
GTR000335343.1	BayCare Cytogenetics Laboratory BayCare Laboratories, LLC	UBE3A	FISH-metaphase	Digital / Virtual karyotyping	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Florida 10D2029560 335343						
GTR000505711.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 505711	UBE3A	Methylation analysis	PCR-RFLP with Southern hybridization	Peripheral (whole) blood	No	No
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|ALDOA|ALDOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CACNA2D4|CASP8|CAT|CBS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM1L|DPM1|DPM3|DSP|DTNBP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCT					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		K|GM2A|GNAT1|GNAT2|GNE|GNPTAB|GNS|GOT1|GPD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|MC1R|MCCC1|MCCC2|MCEE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDU					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		FB3|NDUFS1|NDUFS2|ND UFS3|NDUFS4|NDUFS6|N DUFS7|NDUFS8|NDUFV1| NDUFV2|NEFH|NEUROD1| NFU1|NHS|NME1|NPHP1| NPHP3|NPHP4|NRL|NT5C 3A|NUBPL|NYX|OAT|OCA2 |OCRL|OGDH|OGG1|OPA1 |OPA3|OPN1LW|OPN1MW| OPTN|OSTM1|OTC|OTX2| OXCT1|PAH|PANK2|PARK 2|PARK7|PAX2|PAX6|PC|P CCA|PCCB|PCDH15|PCK2 |PDE6A|PDE6B|PDE6C|PD E6G|PDHA1|PDHB|PDP1|P DSS1|PDSS2|PDZD7|PFK M|PGAM2|PGM1|PHB|PHK A1|PHKA2|PHKB|PHKG2|P HOX2A|PHYH|PINK1|PITP NM3|PITX2|PITX3|PLA2G2 A|PLOD2|PLOD3|PMM2|P NKD|PNPLA2|POLG|POLG 2|PPARG|PPARGC1B|PPI B|PPOX|PPP2R1B|PRCD| PRKCG|PRODH|PROM1|P RPF31|PRPF6|PRPF8|PRP H2|PSAP|PSEN1|PTRF|PT S|PUS1|PYCR1|PYGL|PYG M|QDPR|RAF1|RARS2|RA X|RB1|RD3|RDH12|RDH5| REEP1|RET|RFT1|RGR|R GS9|RHO|RILP|RIMS1|RL BP1|RNASEL|RP1|RP1L1| RP2|RP9|RPE65|RPGR|RP GRIP1|RPL35A|RPS14|RR M2B|RS1|SAG|SARDH|SA RS2|SCO2|SCP2|SDHAF1| SDHAF2|SDHB|SDHC|SEC ISBP2|SEMA4A|SEPT9|SE RPINF1|SGSH|SHH|SIX6|S LC16A1|SLC22A4|SLC22A					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|TRPM1|TSFM|TSPAN12|TTC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512051.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512051	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512052.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512052	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	NR	NR	No
GTR000512053.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512053	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	NR	NR	No
GTR000512054.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512054	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	NR	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512055.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512055	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512056.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512056	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512174.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512174	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512175.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512175	UBE3A	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512176.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512176	UBE3A	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000006790.1	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 6790	15q11.2	FISH-metaphase|Methylation analysis|Uniparental disomy study (UPD)	Other|Methylation-specific PCR|Other	Amniocytes|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000206881.1	Center for Human Genetics, Inc Massachusetts 22D0650242 206881	15q11-q13	FISH-metaphase|Methylation analysis|Sequence analysis of the entire coding region|Uniparental disomy study (UPD)	Fluorescence in situ Hybridization (FISH)|Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis|PCR on ABI	NR	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000196491.3	ChildLab Molecular Genetics Laboratory Nationwide Children's Hospital Ohio 36D0665271 196491	15q11.2-q13	FISH-metaphase|Methylation analysis|Sequence analysis of the entire coding region|Uniparental disomy study (UPD)	FISH for 15q11.2 deletion|Methylation-specific PCR|Bi-directional Sanger Sequence Analysis|characterization of microsatellite polymorphisms on ABI instrument	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000295235.1	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 295235	15q11-q13	FISH-metaphase	Metaphase FISH analysis	Amniocytes|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000005568.2	Clinical Molecular Genetics Laboratory All Children's Hospital Florida 10D0700790 5568	15q11-q13	Methylation analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Buccal swab|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	500883	RSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|BRD2|BTD|BUB1B|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB			Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDH2|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PR				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000500936.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Produ	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	22D2035168 500936	3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI			ct of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	503671	GK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|			(POC)|Saliva|Serum|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PK					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		LR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510912.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 510912	ADSL|ALDH7A1|ARHGEF9|ARX|ATP6AP2|CACNB4|CDKL5|CHD2|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CSTB|CTSD|EFHC1|EPM2A|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GRIN2A|KANSL1|KCNJ10|KCNQ2|KCNQ3|KCNT1|K	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fetal blood|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CTD7|LGI1|LIAS|MAGI2|MBD5|MECP2|MEF2C|MFSD8|NHLRC1|NRXN1|PCDH19|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRRT2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYN1|SYNGAP1|TBC1D24|TCF4|TPP1|TSC1|TSC2|UBE3A|ZEB2					
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000327615.1	Cytogenetics and Molecular Diagnostics Lab CGC Genetics USA New Jersey 31D1085261 327615	SNRPN|UBE3A, 15q11.2	FISH-interphase|FISH-metaphase|Deletion/duplication analysis|Methylation analysis|Sequence analysis of the entire coding region	FISH|FISH|Multiplex Ligation-dependent Probe Amplification (MLPA)|Methylation-specific PCR|Uni-directional Sanger sequencing	Cell-free DNA|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000325589.3	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center Ohio 36D0965797 325589	15q11.2	FISH-metaphase|Methylation analysis	FISH|Methylation-specific PCR	Peripheral (whole) blood	Decline to answer	Yes
GTR000013524.1	Cytogenetics Laboratory Indiana University School of Medicine Indiana 15D0647198 13524	15q11.2-q13	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization (FISH)|Fluorescence in situ hybridization (FISH)	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fibroblasts|Fresh tissue|Peripheral (whole)	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					blood|Product of conception (POC)|Skin|Urine		
GTR000032601.1	Cytogenetics Laboratory SUNY Upstate Medical University New York 33D0654590 32601	15q11.2-q13	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Cord blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin	Yes	Yes
GTR000500042.2	Cytogenetics Laboratory ARUP Laboratories, Inc. Utah 46D0523979 500042	15q11.2-q13|17p11.2|17p13.3|22q11.2|22q13.3|4p16.3|5p15.2|7q11.23|Xp22.3|Yp11.3	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000501320.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501320	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	No	No
GTR000501321.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501321	UBE3A	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512416.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512416	ABAT|ADSL|ALDH7A1|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|BCKDK|CACNA1A|CACNB4|CASK|CASR|CDKL5|CENPJ|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DNAJC5|EFHC1|EMX2|EPM2A|FLNA|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GPR56|GPR98|GRIN2A|HCN1|HCN4|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MTHFR|NDE1|NDUFA1|NHLRC1|NRXN1|OPHN1|PAFAH1B1|PCDH19|PHF6|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRICKLE2|PRRT2|RELN|SCARB2|SCN1A|SCN1B|SCN2A|SCN3A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TPP1|TSC1|TSC2|TSEN54|UBE3A|WDR62|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No
GTR000512586.1	Emory Genetics Laboratory Emory University School of Medicine Georgia	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK16|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|F	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	11D0683478 512586	MR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|HCFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711					
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No
GTR000512608.1	Emory Genetics Laboratory	ACTB|ACTG1|ADSL|AHI1|ALDH7A1|ARFGEF2|ARH	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 512608	GEF9|ARX|ASPM|ATP1A2|ATP6AP2|ATR|ATRX|BCKDK|CACNB4|CASC5|CASK|CC2D2A|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP290|CEP41|CEP63|CHMP1A|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DHCR7|DISP1|DNAJC5|EFHC1|EHMT1|EOMES|EPM2A|EXOSC3|FGF8|FKRP|FKTN|FLNA|FOLR1|FOXG1|FOXH1|GABRA1|GABRG2|GAMT|GATM|GLI2|GOSR2|GPR56|GPR98|GRIN2A|GRIN2B|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KIAA1279|KIF7|LAMC3|LARGE|LARP7|LGI1|LIAS|MAGI2|MAPK10|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MKS1|MYCN|NDE1|NHLRC1|NIN|NODAL|NPHP1|NRXN1|OPHN1|ORC4|ORC6|PAFAH1B1|PCDH19|PCNT|PLCB1|PNKP|PNPO|POC1A|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRRT2|PTCH1|RAB18|RAB3GAP1|RAB3GAP2|RARS2|RBBP8|RELN|RPGRIP1L|RTTN|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TGIF		sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|TMEM138|TMEM216|TMEM237|TMEM67|TPP1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|UBE3A|VLDLR|VRK1|WDR62|ZEB2|ZIC2|ZNF335					
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics California 05D2043189 509399	7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|K		Generation (NGS)/Massively parallel sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000509436.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509436	ABCC8|ACY1|ADAMTSL2|ADSL|AGA|ALDH4A1|ALDH5A1|ALDH7A1|AMT|ANTXR2|ARG1|ARSA|ARSB|ASAH1|ASPA|ATP13A2|BTD|CLN3|CLN5|CLN6|CLN8|COL11A2|COL2A1|CTNS|CTSA|CTSC|CTSD|CTSK|DHCR7|DNAJC5|DPYD|DYM|ETFA|ETFB|ETFDH|FH|FOLR1|FUCA1|GAA|GALC|GALNS|GAMT|GBA|GCDH|GCSH|GLA|GLB1|GLDC|GM2A|GNE|GNPTAB|GNPTG|GNS|GPC3|GUSB|HEXA|HEXB|HGSNAT|HPD|HRAS|IDS|IDUA|L2HGDH|LAMA2|LAMP2|LIPA|LMBRD1|MAN2B1|MANBA|MCOLN1|MFSD8|MOCS1|MOCS2|NAGA|NAGLU|NEU1|NPC1|NPC2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX26|PEX3|PEX5|PEX6|PGK1|PHYH|PPT1|PRODH|P	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SAP|QDPR|RAI1|SGSH|SLC17A5|SLC25A15|SLC46A1|SMPD1|SUMF1|SUOX|TCF4|TPP1					
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509442	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		LE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509443.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics	ALDH7A1|ARX|ATP1A2|CACNA1A|CDKL5|FOLR1|FOXG1|GAMT|KCNQ2|MECP2|PCDH19|PHGDH|PNPO|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	California 05D2043189 509443	POLG|PPT1|SCN1A|SLC2A1|STXBP1		parallel sequencing (MPS)			
GTR000509462.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509462	ARX|ATRX|CAV1|CDKL5|CNTNAP2|FOXG1|MECP2|MED17|MEF2C|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|TRAPPC9|UBE3A|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510915	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACSM3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKDHA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|CLYBL|C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYCTK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510916.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California	ABCD1|ACSL4|AFF2|AGTR2|AIFM1|AP1S2|ARHGEF6|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	05D2043189 510916	5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|EBP|FAAH2|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|GSPT2|HCCS|HDAC8|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|L1CAM|LAMP2|MAGT1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RAB40AL|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SRPX2|SYN1|SYP|TAF1|TIMM8A|TSPAN7|UBE2A|UPF3B|USP9X|WDR13|ZCCHC12|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000006812.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 6812	15q11.2-q13	Methylation analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000006814.1	Genetic Services Laboratory	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	University of Chicago Illinois 14D0917593 6814				swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva		
GTR000500120.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500120	UBE3A	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500149.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500149	15q11-q13	Uniparental disomy study (UPD)	Microsatellite analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500158.1	Genetic Services Laboratory University of Chicago Illinois	MECP2|SLC9A6|TCF4|UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	14D0917593 500158				blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva		
GTR000500159.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500159	15q11-q13	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000501098.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501098	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2	Deletion/duplication analysis	Comparative Genomic Hybridization	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000501110.4	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501110	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000507771.3	Genetic Services Laboratory University of Chicago	ARFGEF2|ASPM|ATR|ATRX|CASC5|CASK|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Illinois 14D0917593 507771	63|FOXG1|MCPH1|MECP2|MED17|NDE1|ORC1|ORC4|ORC6|PCNT|PNKP|RAB3GAP1|RBBP8|SLC25A19|SLC2A1|SLC9A6|STAMBP|STIL|TCF4|TSEN2|TSEN34|TSEN54|UBE3A|WDR62|ZEB2|ZNF335					
GTR000226643.3	Genetics Laboratory University of Oklahoma Health Sciences Center Oklahoma 37D0967945 226643	UBE3A	FISH-metaphase|Methylation analysis|Sequence analysis of the entire coding region	G-banding|Methylation-specific PCR|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000204024.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473 204024	SNRPN|UBE3A, Chromosome 15	Methylation analysis|Sequence analysis of the entire coding region|Uniparental disomy study (UPD)	Pyrosequencing|Bi-directional Sanger Sequence Analysis|Methylation-specific PCR	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin	Decline to answer	Yes
GTR000053112.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 53112	15q11-q13	FISH-interphase|FISH-metaphase|Methylation analysis|Sequence analysis of the entire coding region	Other|Other|Methylation-specific PCR|Methylation-specific PCR	Fetal blood|Peripheral (whole) blood	No	Yes
GTR000327662.1	Human Genetics Laboratory,	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACN	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	Amniocytes|Amniotic fluid|Cord	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 327662	A1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|HPRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2		sequencing (MPS)	blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)		
GTR000331374.1	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 331374	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000500958.4	Mayo Clinic Genetic Testing Laboratories Mayo Clinic	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniotic fluid|Chorionic villi|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Minnesota 24D0404292 500958						
GTR000509438.3	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 509438	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500632.3	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500632	15q11.2	Fluorescence in situ hybridization (FISH)	Other	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Dried blood spot (DBS) card|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)	No	Yes
GTR000084937.2	Molecular Diagnostic Laboratory Barnes Jewish Hospital Missouri 26D0438670 84937	UBE3A	Methylation analysis	PCR-RFLP with Southern hybridization	Peripheral (whole) blood	No	Yes
GTR000248613.1	Molecular Diagnostic Laboratory LabCorp North Carolina 34D0655205 248613	15q11-q13	Methylation analysis	Methylation-specific PCR	Amniotic fluid|Buccal swab|Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500320.1	Molecular Diagnostics Laboratory University of California, San Francisco California 05D1024215 500320	15q11.2	Methylation analysis	Other	Cell culture|Peripheral (whole) blood	No	Yes
GTR000336254.1	Molecular Genetics Diagnostic Laboratory Detroit Medical Center University Laboratories Michigan 23D0717194 336254	UBE3A	Methylation analysis|Sequence analysis of the entire coding region	Methylation-specific PCR|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000193077.1	Molecular Genetics Laboratory Children's Mercy Hospital and Clinics Missouri 26D2046586 193077	15q11.2-q13	Deletion/duplication analysis|Methylation analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)|Methylation-specific PCR	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000204883.3	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio	15q11-q13	Methylation analysis	PCR on sodium bisulfite treated DNA	NR	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	36D0656333 204883						
GTR000500585.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500585	CDKL5	Deletion/duplication analysis|Sequence analysis of the entire coding region	Other|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500696.2	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500696	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000501272.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501272	SLC9A6|SNRPN|UBE3A	Deletion/duplication analysis|Methylation analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Methylation-specific PCR|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501273.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501273	UBE3A	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501274.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501274	UBE3A	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501275.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501275	UBE3A	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501276.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501276	UBE3A	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000503131.2	Natera NR California 05D1082992 503131	15q11-q13|1p36|22q11.2|5p|Chromosome 13|Chromosome 18|Chromosome 21|Human genome|Sex chromosome X	Targeted variant analysis	SNP Detection	Cell-free DNA|Maternal blood sample	Decline to answer	No
GTR000507942.2	Pittsburgh Cytogenetics Laboratory	ABCB7|ABCD1|ACSL4|AFF2|AGTR2|AIFM1|ALAS2|ALG13|AMELX|AMER1|AP1	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniotic fluid|Cell culture|Chorionic villi|Fetal	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	University of Pittsburgh Medical Center Pennsylvania 39D0673863 507942	S2|AR|ARHGEF6|ARHGEF9|ARSE|ARX|ATP2B3|ATP6AP2|ATP7A|ATRX|AVPR2|BCOR|BRWD3|BTK|CACNA1F|CASK|CDKL5|CFP|CHM|CHRDL1|CLCN5|CLIC2|COL4A5|COX7B|CSF2RA|CYBB|DCX|DKC1|DLG3|DMD|EBP|EDA|EFNB1|EMD|F8|F9|FAM58A|FANCB|FHL1|FLNA|FTSJ1|G6PD|GATA1|GDI1|GJB1|GK|GLA|GPR143|GRIA3|HCCS|HCFC1|HDAC8|HPRT1|HSD17B10|IDS|IGBP1|IGSF1|IKBKG|IL1RAPL1|IL2RG|KAL1|KDM5C|KDM6A|L1CAM|LAMP2|MAGT1|MAMLD1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NHS|NLGN3|NLGN4X|NR0B1|NSDHL|OCRL|OFD1|OPHN1|OPN1MW|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PHKA1|PHKA2|PIGA|PLP1|POLA1|PORCN|POU3F4|PQBP1|PRPS1|RAB39B|RAB40AL|RBM10|RP2|RPGR|RPL10|RPS6KA3|RS1|SAT1|SERPINA7|SH2D1A|SHOX|SHROOM4|SLC16A2|SLC35A2|SLC6A14|SLC6A8|SLC9A6|SMPX|SMS|SOX3|SRPX2|SRY|STS|SYN1|SYP|TAF1|TAZ|TBX22|TIMM8A|TMLHE|TRAPPC2|TSPAN7|UBA1|UBE2A|UBQLN2|UPF3B|VMA21|WAS|WDR45|XIAP|XK|ZC4H2|ZDHHC15|ZDHHC9|ZIC3|ZNF41|ZNF674|ZNF711|ZNF8			blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1					
GTR000504348.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504348	15q11.2	FISH-metaphase	Other	Peripheral (whole) blood	No	Yes
GTR000506039.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506039	15q11.2-q13	FISH-metaphase	Other	Amniocytes|Chorionic villi|Peripheral (whole) blood	No	Yes
GTR000501919.2	Transgenomic Transgenomic Connecticut 07D0995237 501919	AARS2|AASS|ABAT|ABCB7|ABCD1|ABHD5|ACAD8|ACAD9|ACADL|ACADM|ACADSB|ACADVL|ACAT1|ACSF3|ACSL4|ADCK3|AFG3L2|AGXT|AIFM1|AIFM2|AK2|AKR1D1|ALAS2|ALDH18A1|ALDH4A1|ALDH5A1|ALDH6A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMACR|AMT|APEX2|APTX|ARG1|ARMS2|ASL|ASS1|ATL1|ATM|ATP5A1|ATP5B|ATP5C1|ATP5D|ATP5E|ATP5F1|ATP5G1|ATP5G2|ATP5G3|ATP5I|ATP5J|ATP5O|ATP7B|ATPAF1|ATPAF2|ATPIF1|ATXN10|ATXN7|AUH|B4GALT1|BCKDHA|BCKDHB|BCS1L|BRAF|BTD|C10orf2|C12orf65|C14orf2|CAPN3|CARS2|CAV3|CDKL5|C	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		HKB|CISD2|CLN3|CLN5|CLN6|CLN8|CMC1|COG1|COG7|COG8|COQ2|COQ3|COQ4|COQ6|COQ7|COQ9|COX10|COX11|COX15|COX17|COX18|COX19|COX4I1|COX4I2|COX5A|COX5B|COX6A1|COX6A2|COX6B1|COX6C|COX7A1|COX7A2|COX7A2L|COX7B|COX7B2|COX7C|COX8A|CPOX|CPS1|CPT1A|CPT2|CRLS1|CRYAB|CTNS|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYC1|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP27A1|CYP27B1|CYP7B1|D2HGDH|DARS2|DBT|DCX|DECR1|DGUOK|DLAT|DLD|DLST|DMGDH|DMPK|DNAJC19|DNM1L|DNM2|DOLK|DPAGT1|DPM1|DPM3|EARS2|ECSIT|EIF2AK3|ELOVL4|ETFA|ETFB|ETFDH|ETHE1|FA2H|FARS2|FASTKD2|FECH|FGF14|FH|FOXG1|FOXRED1|FXN|GAA|GAD1|GAMT|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLA|GLDC|GLRX5|GLUD1|GNPAT|GPD2|GPHN|HADH|HADHA|HADHB|HARS2|HAX1|HCCS|HFE|HK1|HLCS|HMGCL|HMGCS2|HSD17B10|HSD3B2|HSPD1|IARS2|IDH2|ISCU|ITPR1|IVD|KARS|KCNC3|KCNJ11|KIAA0196|KIAA0226|KIF1B|KIF5A|LARS2|LETM1|LMBRD1|LRPPRC|MAOA|MAPT|MARS2|MCCC1|MC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CC2|ME2|MECP2|MFN2|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MPV17|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|MYH7|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA3|NDUFA4|NDUFA5|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAB1|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB1|NDUFB10|NDUFB11|NDUFB2|NDUFB3|NDUFB4|NDUFB5|NDUFB6|NDUFB7|NDUFB8|NDUFB9|NDUFC1|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NEFL|NIPA1|NKX2-1|NPC1|NPC2|NUBPL|OAT|OGDH|OPA1|OPA3|OTC|OXA1L|OXCT1|PAFAH1B1|PANK2|PARL|PARS2|PC|PCCA|PCCB|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX13|PHB|PHYH|PLP1|PMM2|PNKD|PNPLA2|PNPLA3|POLG|POLG2|PPM1B|PPOX|PPT1|PREPL|PRKCG|PRODH|PUS1|PWAR1|RARS2|REEP1|RFT1|RMRP|RNASEH2A|RNASEH2B|RNASEH2C|RRM2B|RYR1|SAMHD1|S					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ARS2|SCN1A|SCO1|SCO2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SGCD|SLC12A3|SLC16A2|SLC19A2|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A16|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC29A3|SLC2A1|SLC2A10|SLC33A1|SLC35A1|SLC35C1|SLC3A1|SLC52A1|SLC6A8|SLC7A9|SOD1|SPAST|SPG11|SPG20|SPG7|SPTBN2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TACO1|TARS2|TAZ|TIMM8A|TK2|TMEM126A|TMEM70|TOP1MT|TPM2|TPP1|TRMU|TSFM|TTBK2|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCR10|UQCR11|UQCRB|UQCRC1|UQCRC2|UQCRFS1|UQCRH|UQCRQ|UROS|USMG5|VARS2|WARS2|WFS1|XDH|XPNPEP3|YARS2|ZFYVE26					
GTR000509336.2	Transgenomic Transgenomic Connecticut 07D0995237 509336	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC3					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000025318.1	United States Air Force, DNA Diagnostic Laboratory United States Air Force Mississippi NR 25318	UBE3A	Methylation analysis	Methylation-specific PCR	Amniocytes|Peripheral (whole) blood	No	No
GTR000305769.2	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305769	15q11-q13	FISH-interphase|FISH-metaphase|Deletion/duplication analysis|Methylation analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray|Methylation-specific PCR	Amniocytes|Amniotic fluid|Cord blood|Peripheral (whole) blood	Decline to answer	Yes

NR – Not reported

Table D-2. Genetic tests for Autism Spectrum Disorder

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322654.3	Ambry Genetics Ambry Genetics California 05D0981414 322654	CDKL5	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Isolated DNA|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000515184.1	Athena Diagnostics Inc Massachusetts 22D0069726 515184	SHANK2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515188.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515188	PTEN	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000507870.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507870	PTEN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000507872.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507872	PTEN	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	Decline to answer	No
GTR000507873.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507873	PTEN	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	Decline to answer	No
GTR000507874.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507874	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000507875.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507875	PTEN	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000507876.1	Baylor Medical	PTEN	Targeted variant analysis	Bi-directional Sanger	Not reported	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratories Baylor College of Medicine Texas 45D0660090 507876			Sequence Analysis		answer	
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|ALDOA|ALDOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CACNA2D4|CASP8|CAT|C	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		BS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM1L|DPM1|DPM3|DSP|DTNBP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCTK|GM2A|GNAT1|GNAT2|GNE|GNPTAB|GNS|GOT1|G					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|MC1R|MCCC1|MCCC2|MCEE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB3|NDUFS1|NDUFS2|ND					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		UFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NEFH|NEUROD1|NFU1|NHS|NME1|NPHP1|NPHP3|NPHP4|NRL|NT5C3A|NUBPL|NYX|OAT|OCA2|OCRL|OGDH|OGG1|OPA1|OPA3|OPN1LW|OPN1MW|OPTN|OSTM1|OTC|OTX2|OXCT1|PAH|PANK2|PARK2|PARK7|PAX2|PAX6|PC|PCCA|PCCB|PCDH15|PCK2|PDE6A|PDE6B|PDE6C|PDE6G|PDHA1|PDHB|PDP1|PDSS1|PDSS2|PDZD7|PFKM|PGAM2|PGM1|PHB|PHKA1|PHKA2|PHKB|PHKG2|PHOX2A|PHYH|PINK1|PITPNM3|PITX2|PITX3|PLA2G2A|PLOD2|PLOD3|PMM2|PNKD|PNPLA2|POLG|POLG2|PPARG|PPARGC1B|PPIB|PPOX|PPP2R1B|PRCD|PRKCG|PRODH|PROM1|PRPF31|PRPF6|PRPF8|PRPH2|PSAP|PSEN1|PTRF|PTS|PUS1|PYCR1|PYGL|PYGM|QDPR|RAF1|RARS2|RAX|RB1|RD3|RDH12|RDH5|REEP1|RET|RFT1|RGR|RGS9|RHO|RILP|RIMS1|RLBP1|RNASEL|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPL35A|RPS14|RRM2B|RS1|SAG|SARDH|SARS2|SCO2|SCP2|SDHAF1|SDHAF2|SDHB|SDHC|SECISBP2|SEMA4A|SEPT9|SERPINF1|SGSH|SHH|SIX6|SLC16A1|SLC22A4|SLC22A					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|TRPM1|TSFM|TSPAN12|TTC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512051.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512051	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512052.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512052	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No
GTR000512053.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512053	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No
GTR000512054.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512054	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512055.1	Baylor Medical	MECP2	Targeted variant analysis	Bi-directional Sanger	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512055			Sequence Analysis			
GTR000512056.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512056	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000243196.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 243196	NLGN3|NLGN4X|SNRPN	FISH-metaphase|Sequence analysis of the entire coding region	Fluorescence in situ Hybridization (FISH)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000311732.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 311732	PTEN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000295241.1	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 295241	Human genome	Detection of homozygosity|Detection of homozygosity	SNP Detection|SNP Detection	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fetal blood|Fibroblasts|Isolated DNA|Product of conception (POC)|Saliva|Skin	Decline to answer	No
GTR000500030.2	Clinical Molecular Genetics Diagnostic Laboratory University of Miami Miller School of Medicine Florida 10D2024894 500030	Human genome	Deletion/duplication analysis	Microarray	Peripheral (whole) blood	Yes (Pre-Test), Decline to Answer (Post-Test)	Yes
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500883	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		BRD2|BTD|BUB1B|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYA					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		L1|IDH2|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNAS					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		EH2A|RNASEH2B|RNASEH2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000500936.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500936	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|H					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		MGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PN					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		MT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 503671	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALD	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMG					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|P					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		OLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000308062.1	DNA Diagnostic Laboratory at Johns Hopkins Johns Hopkins Hospital Maryland 21D0692357 308062	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501369.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501369	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501370.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501370	PTEN	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501803.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501803	NLGN3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501804.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501804	NLGN3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501805.1	Emory Genetics	NLGN4X	Sequence analysis of the	Bi-directional Sanger	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Emory University School of Medicine Georgia 11D0683478 501805		entire coding region	Sequence Analysis			
GTR000501807.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501807	NLGN4X	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501810.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501810	Human genome	Karyotyping	Microarray	Cord blood|Dried blood spot (DBS) card|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000501811.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501811	Human genome	Karyotyping	Microarray	Cord blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood	No	No
GTR000502463.1	Emory Genetics Laboratory	NRXN1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 502463						
GTR000502464.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502464	NRXN1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502824.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502824	RPL10	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502826.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502826	RPL10	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000503014.3	Emory Genetics Laboratory Emory University	Not reported	Analyte	Dimethylene Blue binding Quantitation and Thin Layer Chromatography|GC-MS|Isotope dilution, LC-	Plasma|Urine	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	School of Medicine Georgia 11D0683478 503014			MS/MS|MALDI-TOF/TOF|ion-exchange chromatography			
GTR000503286.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503286	FMR1, Human Genome	Analyte|Karyotyping|Targeted variant analysis	Dimethylene Blue binding Quantitation and Thin Layer Chromatography|GC-MS/MS|Ion-Exchange Chromatography|Isotope dilution method by LC-MS/MS|MALDI-TOF/TOF|Comparative Genomic Hybridization|Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000503288.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503288	FMR1, Human Genome	Karyotyping|Targeted variant analysis	Comparative Genomic Hybridization|Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000512416.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512416	ABAT|ADSL|ALDH7A1|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|BCKDK|CACNA1A|CACNB4|CASK|CASR|CDKL5|CENPJ|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DNAJC5|EFHC1|EMX2|EPM2A|FLNA|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GPR56|GPR9	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		8|GRIN2A|HCN1|HCN4|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MTHFR|NDE1|NDUFA1|NHLRC1|NRXN1|OPHN1|PAFAH1B1|PCDH19|PHF6|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRICKLE2|PRRT2|RELN|SCARB2|SCN1A|SCN1B|SCN2A|SCN3A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TPP1|TSC1|TSC2|TSEN54|UBE3A|WDR62|ZEB2					
GTR000512417.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512417	ADAM17|AICDA|ATG16L1|BTK|C1orf106|CD40LG|COL7A1|CYBA|CYBB|DCLRE1C|DOCK8|ERAP2|FERMT1|FOXP3|FUT2|G6PC3|GUCY2C|HPS1|HPS4|HPS6|ICOS|IL10|IL10RA|IL18RAP|IL23R|IL2RA|IRGM|ITGAM|LRBA|MEFV|MVK|NCF2|NCF4|NOD2|PIK3R1|PLCG2|PTEN|PTPN22|RAC2|RB1|RET|SH2D1A|SLC37A4|STAT1|STXBP2|TTC37|WAS|XIAP	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512586.1	Emory Genetics Laboratory Emory University School of	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Medicine Georgia 11D0683478 512586	6|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|HCFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711					
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ZEB2					
GTR000512606.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512606	CUL4B|EZH2|GLI3|GPC3|MED12|NFIX|NSD1|PHF6|PTCH1|PTEN|UPF3B	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512650.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512650	ALK|APC|ATM|BMPR1A|BRCA1|BRCA2|BRIP1|CDC73|CDH1|CDKN1C|CDKN2A|CHEK2|EPCAM|FH|FLCN|GPC3|MAX|MEN1|MET|MLH1|MSH2|MSH6|MUTYH|NBN|NF2|PALB2|PHOX2B|PMS1|PMS2|PRKAR1A|PTCH1|PTEN|RAD51C|RAD51D|RET|SDHAF2|SDHB|SDHC|SDHD|SMAD4|STK11|SUFU|TMEM127|TP53|VHL|WT1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000507957.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 507957	APC|AXIN2|BMPR1A|BUB1B|CDH1|CDKN2A|CHEK2|EPCAM|EXO1|FLCN|GALNT12|MLH1|MSH2|MSH6|MUTYH|PMS1|PMS2|PTEN|SMAD4|STK11|TP53	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000507985.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California	BRCA1|BRCA2|CDK4|CDKN2A|ERCC3|MC1R|MITF|PTEN|RB1|TERT|TP53|TYR|WRN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	05D2043189 507985						
GTR000508339.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 508339	APC|ATM|ATR|AXIN2|BAP1|BARD1|BLM|BMPR1A|BRCA1|BRCA2|BRIP1|CDH1|CDK4|CDKN2A|CHEK2|CTNNB1|EPCAM|FANCC|HOXB13|MLH1|MRE11A|MSH2|MSH6|MUTYH|NBN|PALB2|PALLD|PMS2|PTEN|RAD50|RAD51|RAD51C|RAD51D|SMAD4|STK11|TP53|VHL|XRCC2|XRCC3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|S	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		OX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6					
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509399	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RN					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000509427.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509427	AIP|APC|ATM|ATR|AXIN2|BAP1|BARD1|BLM|BMPR1A|BRCA1|BRCA2|BRIP1|BUB1B|CDH1|CDK4|CDKN1B|CDKN2A|CHEK2|CTNNB1|CYLD|DDB2|DICER1|EGFR|EGLN1|EPCAM|ERCC2|ERCC3|ERCC4|ERCC5|EXO1|EXT1|EXT2|FANCA|FANCB|FANCC|FANCD2|FANCE|FANCF|FANCG|FANCI|FANCL|FANCM|FH|FLCN|GALNT12|GPC3|HOXB13|HRAS|KIF1B|KIT|MAX|MC1R|MEN1|MET|MITF|MLH1|MPL|MRE11A|MSH2|MS	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H3|MSH6|MUTYH|NBN|NF1|NF2|PALB2|PDGFRA|PICALM|PMS1|PMS2|POLD1|PRKAR1A|PRKDC|PRSS1|PTCH1|PTEN|PTPN11|RAD50|RAD51|RAD51C|RAD51D|RB1|RBBP8|RBM15|RECQL4|RET|ROBO2|SBDS|SDHA|SDHAF2|SDHB|SDHC|SDHD|SLX4|SMAD4|SMARCB1|STK11|SUFU|TERT|TMEM127|TP53|TSC1|TSC2|TSHR|TYR|VHL|WRN|WT1|XPA|XPC|XRCC2|XRCC3					
GTR000509431.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509431	ABAT|ACOX1|ALDH3A2|ARSA|ASPA|CSF1R|DARS2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|FAM126A|GFAP|GJC2|HEPACAM|HSPD1|HTRA1|LMNB1|MLC1|NOTCH3|PLP1|PSAP|PTEN|SCP2|SLC25A12	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509442	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MAN					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		BA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509443.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509443	ALDH7A1|ARX|ATP1A2|CACNA1A|CDKL5|FOLR1|FOXG1|GAMT|KCNQ2|MECP2|PCDH19|PHGDH|PNPO|POLG|PPT1|SCN1A|SLC2A1|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509462.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509462	ARX|ATRX|CAV1|CDKL5|CNTNAP2|FOXG1|MECP2|MED17|MEF2C|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|TRAPPC9|UBE3A|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510869.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California	BRAF|HRAS|KRAS|MUTYH|NRAS|PTEN|RET|SDHB|SDHD|TP53	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	05D2043189 510869						
GTR000510914.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510914	BAP1|EPCAM|FH|FLCN|MET|MITF|MLH1|MSH2|MSH6|PMS2|PTEN|SDHA|SDHB|SDHC|SDHD|TP53|TSC1|TSC2|VHL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510915	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACSM3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKDHA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		LN5|CLN6|CLN8|CLYBL|CNR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYCTK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|K					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510916.8	Fulgent Clinical Diagnostics Lab	ABCD1|ACSL4|AFF2|AGTR2|AIFM1|AP1S2|ARHGEF	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Buccal swab|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 510916	6|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|EBP|FAAH2|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|GSPT2|HCCS|HDAC8|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|L1CAM|LAMP2|MAGT1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RAB40AL|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SRPX2|SYN1|SYP|TAF1|TIMM8A|TSPAN7|UBE2A|UPF3B|USP9X|WDR13|ZCCHC12|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	(whole) blood|Saliva		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 327662	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|HPRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No
GTR000515046.3	Illumina Clinical Services Laboratory Illumina California 05D1092911 515046	1,578 genes and variants	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA/Peripheral (whole) blood	Not reported	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501388.2	Lineagen, Inc — Utah 46D2042721 501388	Human genome	Deletion/duplication analysis	Microarray	Buccal swab|Peripheral (whole) blood	No	Yes
GTR000331296.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 331296	CDKL5|MBD5|NLGN3|NLGN4X|SHANK2|SHANK3|SLC9A6|TCF4|UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Isolated DNA	Decline to answer	No
GTR000332993.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 332993	MECP2|PTEN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Isolated DNA	Decline to answer	No
GTR000508887.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 508887	SHANK3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000508888.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 508888	Human genome	Deletion/duplication analysis	Microarray|Real Time Quantitative PCR	Peripheral (whole) blood	Decline to answer	Yes
GTR000509356.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 509356	CDKL5	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000509437.3	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 509437	MEF2C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000509438.3	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 509438	UBE3A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000509439.3	Michigan Medical	SHANK2	Sequence analysis of the	Bi-directional Sanger	Peripheral (whole)	Decline to	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratories University of Michigan Michigan 23D0366712 509439		entire coding region	Sequence Analysis	blood	answer	
GTR000500624.2	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500624	FMR1	Targeted variant analysis	PCR with allele specific hybridization	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Chorionic villi|Cord blood|Dried blood spot (DBS) card|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood|Product of conception (POC)|Saliva|Serum|Skin	No	No
GTR000500625.3	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500625	Human genome	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Chorionic villi|Cord blood|Dried blood spot (DBS) card|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood|Product of	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					conception (POC)|Saliva|Serum|Skin		
GTR000500546.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500546	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500741.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500741	PTEN	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Peripheral (whole) blood	Decline to answer	No
GTR000500742.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500742	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500755.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500755	PTEN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Other|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000503364.3	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503364	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000503365.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503365	PTEN	Deletion/duplication analysis	Microarray	Not reported	Decline to answer	No
GTR000511640.1	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio	PTEN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	36D0656333 511640						
GTR000500513.1	Mount Sinai Genetic Testing Laboratory Icahn School of Medicine at Mount Sinai New York 33D0653419 500513	Human genome	Karyotyping	Microarray	Fetal blood|Fibroblasts|Fresh tissue|Isolated DNA|Please visit http://www.mssm.edu/geneticstesting for test specifi|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin	Yes	Yes
GTR000507942.2	Pittsburgh Cytogenetics Laboratory University of Pittsburgh Medical Center Pennsylvania 39D0673863 507942	ABCB7|ABCD1|ACSL4|AFF2|AGTR2|AIFM1|ALAS2|ALG13|AMELX|AMER1|AP1S2|AR|ARHGEF6|ARHGEF9|ARSE|ARX|ATP2B3|ATP6AP2|ATP7A|ATRX|AVPR2|BCOR|BRWD3|BTK|CACNA1F|CASK|CDKL5|CFP|CHM|CHRDL1|CLCN5|CLIC2|COL4A5|COX7B|CSF2RA|CYBB|DCX|DKC1|DLG3|DMD|EBP|EDA|EFNB1|EMD|F8|F9|FAM58A|FANCB|FHL1|FLNA|FTSJ1|G6PD|GATA1|GDI1|GJB1|GK|GLA|GPR143|GRIA3|HCCS|HCFC1|HDAC8|HPRT1|HSD17B10|IDS|IGBP1|IGSF1|IKBKG|IL1RAPL1|IL2RG|KAL1|KDM5C|KDM6A|L1CAM|LAMP2|MAGT1|MAMLD1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NHS|NLGN3|NLGN4X|NR0B1|NSDHL|OCRL|OFD1|OPH	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniotic fluid|Cell culture|Chorionic villi|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		N1|OPN1MW|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PHKA1|PHKA2|PIGA|PLP1|POLA1|PORCN|POU3F4|PQBP1|PRPS1|RAB39B|RAB40AL|RBM10|RP2|RPGR|RPL10|RPS6KA3|RS1|SAT1|SERPINA7|SH2D1A|SHOX|SHROOM4|SLC16A2|SLC35A2|SLC6A14|SLC6A8|SLC9A6|SMPX|SMS|SOX3|SRPX2|SRY|STS|SYN1|SYP|TAF1|TAZ|TBX22|TIMM8A|TMLHE|TRAPPC2|TSPAN7|UBA1|UBE2A|UBQLN2|UPF3B|VMA21|WAS|WDR45|XIAP|XK|ZC4H2|ZDHHC15|ZDHHC9|ZIC3|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000504341.2	Quest Diagnostics Nichols Institute Chantilly — Virginia 49D0221801 504341	Human genome	Deletion/duplication analysis	Microarray|SNP Detection	Cord blood|Peripheral (whole) blood	No	Yes
GTR000504346.1	Quest Diagnostics Nichols Institute Chantilly — Virginia 49D0221801 504346	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000504349.1	Quest Diagnostics Nichols Institute Chantilly — Virginia 49D0221801 504349	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes
GTR000506028.2	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 506028	Human genome	Deletion/duplication analysis	Microarray|SNP Detection	Cord blood|Peripheral (whole) blood	No	Yes
GTR000506033.2	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 506033	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000506036.2	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 506036	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes
GTR000506042.1	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 506042	15q11-q13	FISH-interphase	Other	Peripheral (whole) blood	No	Yes
GTR000335197.2	Transgenomic Transgenomic Connecticut 07D0995237 335197	PTEN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Fresh tissue|Frozen tissue|Paraffin block	Decline to answer	Yes
GTR000501919.2	Transgenomic Transgenomic Connecticut 07D0995237 501919	AARS2|AASS|ABAT|ABCB7|ABCD1|ABHD5|ACAD8|ACAD9|ACADL|ACADM|ACADSB|ACADVL|ACAT1|ACSF3|ACSL4|ADCK3|AFG3L2|AGXT|AIFM1|AIFM2|AK2|AKR1D1|ALAS2|ALDH18A1|ALDH4A1|ALDH5A1|ALDH6A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMACR|AMT|APEX2|APTX|ARG1|ARMS2|ASL|ASS1|AT	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		L1|ATM|ATP5A1|ATP5B|ATP5C1|ATP5D|ATP5E|ATP5F1|ATP5G1|ATP5G2|ATP5G3|ATP5I|ATP5J|ATP5O|ATP7B|ATPAF1|ATPAF2|ATPIF1|ATXN10|ATXN7|AUH|B4GALT1|BCKDHA|BCKDHB|BCS1L|BRAF|BTD|C10orf2|C12orf65|C14orf2|CAPN3|CARS2|CAV3|CDKL5|CHKB|CISD2|CLN3|CLN5|CLN6|CLN8|CMC1|COG1|COG7|COG8|COQ2|COQ3|COQ4|COQ6|COQ7|COQ9|COX10|COX11|COX15|COX17|COX18|COX19|COX4I1|COX4I2|COX5A|COX5B|COX6A1|COX6A2|COX6B1|COX6C|COX7A1|COX7A2|COX7A2L|COX7B|COX7B2|COX7C|COX8A|CPOX|CPS1|CPT1A|CPT2|CRLS1|CRYAB|CTNS|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYC1|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP27A1|CYP27B1|CYP7B1|D2HGDH|DARS2|DBT|DCX|DECR1|DGUOK|DLAT|DLD|DLST|DMGDH|DMPK|DNAJC19|DNM1L|DNM2|DOLK|DPAGT1|DPM1|DPM3|EARS2|ECSIT|EIF2AK3|ELOVL4|ETFA|ETFB|ETFDH|ETHE1|FA2H|FARS2|FASTKD2|FECH|FGF14|FH|FOXG1|FOXRED1|FXN|GAA|GAD1|GAMT|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLA|GLDC|GLRX					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5|GLUD1|GNPAT|GPD2|GPHN|HADH|HADHA|HADHB|HARS2|HAX1|HCCS|HFE|HK1|HLCS|HMGCL|HMGCS2|HSD17B10|HSD3B2|HSPD1|IARS2|IDH2|ISCU|ITPR1|IVD|KARS|KCNC3|KCNJ11|KIAA0196|KIAA0226|KIF1B|KIF5A|LARS2|LETM1|LMBRD1|LRPPRC|MAOA|MAPT|MARS2|MCCC1|MCCC2|ME2|MECP2|MFN2|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MPV17|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|MYH7|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA3|NDUFA4|NDUFA5|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAB1|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB1|NDUFB10|NDUFB11|NDUFB2|NDUFB3|NDUFB4|NDUFB5|NDUFB6|NDUFB7|NDUFB8|NDUFB9|NDUFC1|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NEFL|NIPA1|NKX2-1|NPC1|NPC2|NUBPL|OAT|OGDH|OPA1|OPA3|OTC|OXA1L|OXCT1|PAFAH1B1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|PANK2|PARL|PARS2|PC|PCCA|PCCB|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX13|PHB|PHYH|PLP1|PMM2|PNKD|PNPLA2|PNPLA3|POLG|POLG2|PPM1B|PPOX|PPT1|PREPL|PRKCG|PRODH|PUS1|PWAR1|RARS2|REEP1|RFT1|RMRP|RNASEH2A|RNASEH2B|RNASEH2C|RRM2B|RYR1|SAMHD1|SARS2|SCN1A|SCO1|SCO2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SGCD|SLC12A3|SLC16A2|SLC19A2|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A16|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC29A3|SLC2A1|SLC2A10|SLC33A1|SLC35A1|SLC35C1|SLC3A1|SLC52A1|SLC6A8|SLC7A9|SOD1|SPAST|SPG11|SPG20|SPG7|SPTBN2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TACO1|TARS2|TAZ|TIMM8A|TK2|TMEM126A|TMEM70|TOP1MT|TPM2|TPP1|TRMU|TSFM|TTBK2|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCR10|UQCR11|UQCRB|UQCRC1|UQCRC2|UQCRFS1|UQCRH|UQCRQ|UROS|USMG5|VARS2|WARS2|WFS1|XDH|XPNPEP3|YARS2|ZFYVE26					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509336.2	Transgenomic Transgenomic Connecticut 07D0995237 509336	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKL					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		E2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000305775.2	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305775	Sex chromosome X	FISH-interphase|FISH-metaphase|Deletion/duplication analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray	Peripheral (whole) blood	Decline to answer	Yes

Table D-3. Genetic tests for Developmental Delay

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000323171.3	(AC) Atlanta Center: Medical Neurogenetics, LLC. Georgia 11D0703390 323171	GFER	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Skin|White blood cell prep	Decline to answer	Yes
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|ALDOA|ALDOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CA	Deletion/duplication analysis	Comparative Genomic Hybridization	—	Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CNA2D4|CASP8|CAT|CBS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM1L|DPM1|DPM3|DSP|DTNBP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCTK|GM2A|GNAT1|GNAT2|GNE|GNPT					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		AB|GNS|GOT1|GPD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|MC1R|MCCC1|MCCC2|MCEE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB3|NDUFS1|N					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		DUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NEFH|NEUROD1|NFU1|NHS|NME1|NPHP1|NPHP3|NPHP4|NRL|NT5C3A|NUBPL|NYX|OAT|OCA2|OCRL|OGDH|OGG1|OPA1|OPA3|OPN1LW|OPN1MW|OPTN|OSTM1|OTC|OTX2|OXCT1|PAH|PANK2|PARK2|PARK7|PAX2|PAX6|PC|PCCA|PCCB|PCDH15|PCK2|PDE6A|PDE6B|PDE6C|PDE6G|PDHA1|PDHB|PDP1|PDSS1|PDSS2|PDZD7|PFKM|PGAM2|PGM1|PHB|PHKA1|PHKA2|PHKB|PHKG2|PHOX2A|PHYH|PINK1|PITPNM3|PITX2|PITX3|PLA2G2A|PLOD2|PLOD3|PMM2|PNKD|PNPLA2|POLG|POLG2|PPARG|PPARGC1B|PPIB|PPOX|PPP2R1B|PRCD|PRKCG|PRODH|PROM1|PRPF31|PRPF6|PRPF8|PRPH2|PSAP|PSEN1|PTRF|PTS|PUS1|PYCR1|PYGL|PYGM|QDPR|RAF1|RARS2|RAX|RB1|RD3|RDH12|RDH5|REEP1|RET|RFT1|RGR|RGS9|RHO|RILP|RIMS1|RLBP1|RNASEL|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPL35A|RPS14|RRM2B|RS1|SAG|SARDH|SARS2|SCO2|SCP2|SDHAF1|SDHAF2|SDHB|SDHC|SECISBP2|SEMA4A|SEPT9|SERPINF1|SGSH|SHH|SIX6|SLC16A1|SLC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		22A4|SLC22A5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|TRPM1|TSFM|TSPAN12|TTC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513					
GTR000295241.1	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's	Human genome	Detection of homozygosity|Detection of homozygosity	SNP Detection|SNP Detection	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fetal	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Hospital Medical Center Ohio 36D0656333 295241				blood|Fibroblasts|Isolated DNA|Product of conception (POC)|Saliva|Skin		
GTR000500252.2	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 500252	Human genome	Deletion/duplication analysis	Microarray	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000500030.2	Clinical Molecular Genetics Diagnostic Laboratory University of Miami Miller School of Medicine Florida 10D2024894 500030	Human genome	Deletion/duplication analysis	Microarray	Peripheral (whole) blood	Yes (Pre-test), Decline to Answer (Post-test)	Yes
GTR000500936.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Seru	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	500936	2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HAD			m|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	22D2035168 503671	GK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HA			(POC)|Saliva|Serum|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		DH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500620.3	Cytogenetics and Microarray Laboratory Kennedy Krieger Institute Maryland 21D0649789 500620	Human genome	Deletion/duplication analysis	Microarray	Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000316021.1	Cytogenetics and Molecular Pathology Laboratory DLP Marquette General Hospital, LLC Michigan 23D1062707 316021	CTNND2	FISH-interphase|FISH-metaphase|Karyotyping	Fluorescent in situ Hybridization|Fluorescent in situ Hybridization|G-banding	Buccal swab|Cell culture|Cord blood|Fetal blood|Peripheral (whole) blood	Yes	No
GTR000167401.2	DNA Diagnostic Laboratory at Johns Hopkins Johns Hopkins Hospital Maryland 21D0692357 167401	FGFR3	Sequence analysis of select exons	Bi-directional Sanger Sequence Analysis	—	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501810.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501810	Human genome	Karyotyping	Microarray	Cord blood|Dried blood spot (DBS) card|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000501811.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501811	Human genome	Karyotyping	Microarray	Cord blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood	No	No
GTR000509392.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509392	ABCC8|ALMS1|ARL6|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BDNF|CEL|CEP290|EIF2AK3|FOXP3|GCK|GLIS3|GNAS|HNF1A|HNF1B|HNF4A|INS|KCNJ11|LEP|LEPR|MAGEL2|MC4R|MKKS|MKS1|NEUROD1|NEUROG3|NTRK2|PCSK1|PDX1|POMC|PTF1A|RFX6|SDCCAG8|SIM1|TRIM32|TTC8|WDPCP|WFS1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	—	Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	509442	1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510915	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACSM3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|A	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKDHA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|CLYBL|CNR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		TK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|ND					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		UFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000515046.3	Illumina Clinical Services Laboratory Illumina California 05D1092911 515046	—	—	—	—	Not reported	Not reported
GTR000510735.1	Insight Medical Genetics Insight Medical Genetics Illinois 14D1000956 510735	ALPL|COL2A1|ESCO2|FGFR2|FGFR3|ROR2|SLC26A2|SOX9	Mutation scanning of select exons	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	No
GTR000501388.2	Lineagen, Inc Utah 46D2042721 501388	Human genome	Deletion/duplication analysis	Microarray	Buccal swab|Peripheral (whole) blood	No	Yes
GTR000500624.2	Michigan State University Clinical Genetics Laboratory Michigan State University	FMR1	Targeted variant analysis	PCR with allele specific hybridization	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Chorionic villi|Cord blood|Dried blood	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Michigan 23D0650879 500624				spot (DBS) card|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood|Product of conception (POC)|Saliva|Serum|Skin		
GTR000500513.1	Mount Sinai Genetic Testing Laboratory Icahn School of Medicine at Mount Sinai New York 33D0653419 500513	Human genome	Karyotyping	Microarray	Fetal blood|Fibroblasts|Fresh tissue|Isolated DNA|Please visit http://www.mssm.edu/genetictesting for test specifi|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin	Yes (Pre-test), Decline to Answer (Post-Test)	Yes
GTR000504341.2	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504341	Human genome	Deletion/duplication analysis	Microarray|SNP Detection	Cord blood|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000504346.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504346	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes
GTR000504349.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504349	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes
GTR000506028.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506028	Human genome	Deletion/duplication analysis	Microarray|SNP Detection	Cord blood|Peripheral (whole) blood	No	Yes
GTR000506033.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506033	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000506036.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506036	Human genome	Karyotyping|Deletion/duplication analysis	G-banding|Microarray|SNP Detection	Peripheral (whole) blood	No	Yes
GTR000501919.2	Transgenomic Transgenomic Connecticut 07D0995237 501919	AARS2|AASS|ABAT|ABCB7|ABCD1|ABHD5|ACAD8|ACAD9|ACADL|ACADM|ACADSB|ACADVL|ACAT1|ACSF3|ACSL4|ADCK3|AFG3L2|AGXT|AIFM1|AIFM2|AK2|AKR1D1|ALAS2|ALDH18A1|ALDH4A1|ALDH5A1|ALDH6A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMACR|AMT|APEX2|APTX|ARG1|ARMS2|ASL|ASS1|ATL1|ATM|ATP5A1|ATP5B|ATP5C1|ATP5D|ATP5E|ATP5F1|ATP5G1|ATP5G2|ATP5G3|ATP5I|ATP5J|ATP5O|ATP7B|ATPAF1|ATPAF2|ATPIF1|ATXN10|ATXN7|AUH|B4GALT1|BCKDHA|BCKDHB|BCS1L|BRAF|BTD|C10orf2|C12orf65|C14orf2|CAPN3|CARS2|CAV3|CDKL5|CHKB|CISD2|CLN3|CLN5|CLN6|CLN8|CMC1|COG1|COG7|COG8|COQ2|COQ3|COQ4|COQ6|COQ7|COQ9|COX10|COX11|COX15|COX17|COX18|COX19|COX4I1|COX4I2|COX5A|COX5B|COX6A1|COX6A2|COX6B1|C	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		OX6C|COX7A1|COX7A2|COX7A2L|COX7B|COX7B2|COX7C|COX8A|CPOX|CPS1|CPT1A|CPT2|CRLS1|CRYAB|CTNS|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYC1|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP27A1|CYP27B1|CYP7B1|D2HGDH|DARS2|DBT|DCX|DECR1|DGUOK|DLAT|DLD|DLST|DMGDH|DMPK|DNAJC19|DNM1L|DNM2|DOLK|DPAGT1|DPM1|DPM3|EARS2|ECSIT|EIF2AK3|ELOVL4|ETFA|ETFB|ETFDH|ETHE1|FA2H|FARS2|FASTKD2|FECH|FGF14|FH|FOXG1|FOXRED1|FXN|GAA|GAD1|GAMT|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLA|GLDC|GLRX5|GLUD1|GNPAT|GPD2|GPHN|HADH|HADHA|HADHB|HARS2|HAX1|HCCS|HFE|HK1|HLCS|HMGCL|HMGCS2|HSD17B10|HSD3B2|HSPD1|IARS2|IDH2|ISCU|ITPR1|IVD|KARS|KCNC3|KCNJ11|KIAA0196|KIAA0226|KIF1B|KIF5A|LARS2|LETM1|LMBRD1|LRPPRC|MAOA|MAPT|MARS2|MCCC1|MCCC2|ME2|MECP2|MFN2|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MPV17|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTR					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		R|MUT|MUTYH|MYH7|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA3|NDUFA4|NDUFA5|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAB1|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB1|NDUFB10|NDUFB11|NDUFB2|NDUFB3|NDUFB4|NDUFB5|NDUFB6|NDUFB7|NDUFB8|NDUFB9|NDUFC1|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NEFL|NIPA1|NKX2-1|NPC1|NPC2|NUBPL|OAT|OGDH|OPA1|OPA3|OTC|OXA1L|OXCT1|PAFAH1B1|PANK2|PARL|PARS2|PC|PCCA|PCCB|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX13|PHB|PHYH|PLP1|PMM2|PNKD|PNPLA2|PNPLA3|POLG|POLG2|PPM1B|PPOX|PPT1|PREPL|PRKCG|PRODH|PUS1|PWAR1|RARS2|REEP1|RFT1|RMRP|RNASEH2A|RNASEH2B|RNASEH2C|RRM2B|RYR1|SAMHD1|SARS2|SCN1A|SCO1|SCO2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SGCD|SLC12A3|SLC16A2|SLC19A2|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A16|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Gene	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC29A3|SLC2A1|SLC2A10|SLC33A1|SLC35A1|SLC35C1|SLC3A1|SLC52A1|SLC6A8|SLC7A9|SOD1|SPAST|SPG11|SPG20|SPG7|SPTBN2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TACO1|TARS2|TAZ|TIMM8A|TK2|TMEM126A|TMEM70|TOP1MT|TPM2|TPP1|TRMU|TSFM|TTBK2|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCR10|UQCR11|UQCRB|UQCRC1|UQCRC2|UQCRFS1|UQCRH|UQCRQ|UROS|USMG5|VARS2|WARS2|WFS1|XDH|XPNPEP3|YARS2|ZFYVE26					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322598.2	Ambry Genetics Ambry Genetics California 05D0981414 322598	FMR1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood	Decline to answer (pre-test), No (post-test)	Yes
GTR000500911.4	Asuragen Clinical Laboratory Asuragen, Inc Texas 45D1069375 500911	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Buccal swab|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000510864.2	Asuragen Clinical Laboratory Asuragen, Inc Texas 45D1069375 510864	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Buccal swab|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000505885.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 505885	FMR1	Methylation analysis	PCR-RFLP with Southern hybridization	Peripheral (whole) blood	No	No
GTR000511954.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	45D0660090 511954						
GTR000511955.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511955	FMR1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000511956.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511956	FMR1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000511957.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511957	FMR1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000511965.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	511965						
GTR000502891.1	Center for Genetics at Saint Francis Saint Francis Hospital Oklahoma 37D0474681 502891	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Fetal blood|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000006798.1	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 6798	FMR1	Targeted variant analysis	PCR-RFLP with Southern hybridization	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000004006.1	Center for Human Genetics, Inc Massachusetts 22D0650242 4006	FMR1	Methylation analysis|Targeted variant analysis	Methylation-specific PCR|Trinucleotide repeat by PCR or Southern Blot	Not reported	Decline to answer	No
GTR000004082.1	Clinical Molecular Genetics Laboratory All Children's Hospital Florida	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Buccal swab|Chorionic villi|Fibroblasts|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	10D0700790 4082						
GTR000501051.2	Counsyl California 05D1102604 501051	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SH	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000503904.1	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center Ohio 36D0965797 503904	FMR1	Microsatellite instability testing (MSI)	Trinucleotide repeat by PCR or Southern Blot	Peripheral (whole) blood	Decline to answer	Yes
GTR000501519.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501519	FMR1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501520.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501520	FMR1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501753.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501753	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000503266.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503266	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000503286.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503286	FMR1, Human Genome	Analyte|Karyotyping|Targeted variant analysis	Dimethylene Blue binding Quantitation and Thin Layer Chromatography|GC-MS/MS|Ion-Exchange Chromatography|Isotope dilution method by LC-MS/MS|MALDI-TOF/TOF|Comparative Genomic Hybridization|Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000503288.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478	FMR1, Human Genome	Karyotyping|Targeted variant analysis	Comparative Genomic Hybridization|Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	503288						
GTR000512586.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512586	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK16|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|HCFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		FAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2					
GTR000500301.3	Fragile X Laboratory/IBR Specialty Clinical Laboratories New York State Institute for Basic Research in Developmental Disabilities New York 33D0860102 500301	FMR1	Targeted variant analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Dried blood spot (DBS) card|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|White blood cell prep	No	No
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCD	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6					
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509442	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EB	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab/Isolated DNA/Peripheral (whole) blood/Saliva	Decline to answer	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|N					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|US					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000510916.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510916	ABCD1|ACSL4|AFF2|AGTR2|AIFM1|AP1S2|ARHGEF6|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|EBP|FAAH2|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|GSPT2|HCCS|HDAC8|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|L1CAM|LAMP2|MAGT1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RAB40AL|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SRPX2|SYN1|SYP|TAF1|TIMM8A|TSPAN7|UBE2A|UPF3B|USP9X|WDR13|ZCCHC12|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511180.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 511180	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Buccal swab|Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000226645.1	Genetics Laboratory University of Oklahoma Health Sciences Center Oklahoma 37D0967945 226645	FMR1	Targeted variant analysis	PCR-RFLP with Southern hybridization	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000500314.1	Genetics Laboratory Shodair Children's Hospital Montana 27D0652530 500314	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000500315.2	Genetics Laboratory Shodair Children's Hospital Montana 27D0652530 500315	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000004034.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473 4034	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin	Decline to answer	Yes
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 327662	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|HPRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000004902.1	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 4902	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000331372.1	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 331372	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000005424.4	Molecular and Biochemical Genetics Laboratory Dayton Children's Hospital Ohio 36D0859317 5424	FMR1	Targeted variant analysis	Southern Blot and PCR	Isolated DNA|Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000005238.1	Molecular Diagnostic Laboratory Barnes Jewish Hospital Missouri 26D0438670 5238	FMR1	Targeted variant analysis	PCR with Fragment Analysis|PCR-RFLP with Southern hybridization	Peripheral (whole) blood	Decline to answer	Yes
GTR000026021.3	Molecular Diagnostic Laboratory Nebraska Medical Center Nebraska 28D0453728 26021	FMR1|POF1B	Methylation analysis|Targeted variant analysis	PCR-RFLP with Southern hybridization|Trinucleotide repeat by PCR or Southern Blot	Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000500327.1	Molecular Diagnostics Laboratory University of California, San Francisco California 05D1024215 500327		Methylation analysis|Targeted variant analysis	Other|Trinucleotide repeat by PCR or Southern Blot	Cell culture|Peripheral (whole) blood	No	Yes
GTR000500334.1	Molecular Diagnostics Laboratory University of California, San Francisco California 05D1024215 500334	FMR1	Methylation analysis|Targeted variant analysis	Other|Trinucleotide repeat by PCR or Southern Blot	Cell culture|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000005250.2	Molecular Genetics Diagnostic Laboratory Detroit Medical Center University Laboratories Michigan 23D0717194 5250	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Peripheral (whole) blood	Decline to answer	Yes
GTR000006804.3	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 6804	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Decline to answer	Yes
GTR000203713.1	Molecular Genetics Laboratory Children's Mercy Hospital and Clinics Missouri 26D2046586 203713	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000501532.1	Molecular Genetics Laboratory Children's Hospital of	FMR1	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Philadelphia Pennsylvania 39D0198678 501532				(whole) blood		
GTR000502738.3	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 502738	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No
GTR000502739.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 502739	FMR1	Deletion/duplication analysis	Microarray	Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	No	No
GTR000512245.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 512245	FMR1	Methylation analysis|Targeted variant analysis	PCR/Capillary Electrophoresis. Methylation analysis is performed to distinguish between premutation and full mutation alleles.|PCR/Capillary Electrophoresis. Methylation analysis is performed to distinguish between premutation and full mutation alleles.	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512246.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 512246	FMR1	Methylation analysis|Targeted variant analysis	PCR/Capillary Electrophoresis. Methylation analysis is performed to distinguish between premutation and full mutation alleles.| PCR/Capillary Electrophoresis. Methylation analysis is performed to distinguish between premutation and full mutation alleles.	Amniocytes|Amniotic fluid|Cord blood|Peripheral (whole) blood	Decline to answer	Yes
GTR000004062.1	Molecular Pathology Laboratory Ohio State University Ohio 36D1080238 4062	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Peripheral (whole) blood	Decline to answer	Yes
GTR000511497.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 511497	FMR1	Methylation analysis|Microsatellite instability testing (MSI)	Methylation-specific PCR|Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000508090.1	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 508090	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	No	Yes
GTR000508091.1	Quest Diagnostics Nichols Institute San Juan Capistrano — California 05D0643352 508091	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	No	Yes
GTR000025286.1	United States Air Force, DNA Diagnostic Laboratory United States Air Force Mississippi — 25286	FMR1	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Peripheral (whole) blood	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000219909.1	University of Iowa Diagnostic Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 219909	FMR1	Methylation analysis|Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot|Trinucleotide repeat by PCR or Southern Blot	Cell culture|Peripheral (whole) blood	Decline to answer	Yes
GTR000305777.1	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305777	FMR1	Methylation analysis|Targeted variant analysis	PCR-RFLP with Southern hybridization|PCR with allele specific hybridization	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes

Table D-5. Genetic tests for Intellectual Disability

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000326450.5	(AC) Atlanta Center: Medical Neurogenetics, LLC. — Georgia 11D0703390 326450	FKTN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Skin|White blood cell prep	Decline to answer	Yes
GTR000326455.5	(AC) Atlanta Center: Medical Neurogenetics, LLC. — Georgia 11D0703390 326455	FKRP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Skin|White blood cell prep	Decline to answer	Yes
GTR000322643.1	Ambry Genetics Ambry Genetics California 05D0981414 322643	BRWD3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322644.1	Ambry Genetics Ambry Genetics California 05D0981414 322644	ACSL4	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322645.1	Ambry Genetics Ambry Genetics California 05D0981414 322645	AGTR2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322646.1	Ambry Genetics Ambry Genetics California 05D0981414 322646	AP1S2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322647.1	Ambry Genetics Ambry Genetics California 05D0981414 322647	ARHGEF6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322650.1	Ambry Genetics Ambry Genetics California 05D0981414 322650	DLG3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322651.1	Ambry Genetics Ambry Genetics California 05D0981414 322651	FTSJ1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322652.1	Ambry Genetics Ambry Genetics California 05D0981414 322652	GDI1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322653.1	Ambry Genetics Ambry Genetics California 05D0981414 322653	IL1RAPL1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322655.1	Ambry Genetics Ambry Genetics California 05D0981414 322655	PAK3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322658.1	Ambry Genetics Ambry Genetics California 05D0981414 322658	TSPAN7	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322659.3	Ambry Genetics Ambry Genetics California 05D0981414 322659	UPF3B	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000322660.3	Ambry Genetics Ambry Genetics California 05D0981414 322660	ZDHHC9	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000322661.1	Ambry Genetics Ambry Genetics California 05D0981414 322661	ZNF41	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322666.5	Ambry Genetics Ambry Genetics California 05D0981414 322666	CASK	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000322667.1	Ambry Genetics Ambry Genetics California 05D0981414 322667	KDM5C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322668.2	Ambry Genetics Ambry Genetics California 05D0981414 322668	SYP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322669.1	Ambry Genetics Ambry Genetics California 05D0981414 322669	ZNF711	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322670.1	Ambry Genetics Ambry Genetics California 05D0981414 322670	ZNF81	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000322720.4	Ambry Genetics Ambry Genetics California 05D0981414 322720	CUL4B	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000322721.3	Ambry Genetics Ambry Genetics California 05D0981414 322721	FGD1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000322722.5	Ambry Genetics Ambry Genetics California 05D0981414 322722	KIAA2022	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000322723.1	Ambry Genetics Ambry Genetics California 05D0981414 322723	ZNF674	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000512790.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512790	ARHGEF9|ARX|ATP6AP2|ATRX|CASK|CDKL5|CUL4B|DCX|FGD1|GPC3|GRIA3|HSD17B10|KDM5C|MECP2|OFD1|OPHN1|PAK3|PCDH19|PHF6|PLP1|PQBP1|RAB39B|SLC9A6|SMC1A|SMS|SRPX2|SYP	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000512791.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512791	ARHGEF9|ARX|CDKL5|CNTNAP2|FOXG1|GABRG2|GRIN2A|KCNT1|MECP2|NRXN1|PCDH19|PNKP|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYNGAP1|TCF4|TREX1|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512792.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512792	ARX|CDKL5|FOXG1|GABRB3|GRIN2A|MEF2C|SCN2A|SLC25A22|SPTAN1|STXBP1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000512794.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512794	ALDH7A1|CACNA1A|CASR|CHRNA2|CHRNA4|CHRNB2|CSTB|DEPDC5|EFHC1|EPM2A|GABRA1|GABRB3|GABRD|GABRG2|GRIN2A|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|MBD5|ME2|NHLRC1|PCDH19|PRICKLE1|PRICKLE2|PRRT2|SCARB2|SCN1A|SCN1B|SCN2A|SCN9A|SLC2A1|SLC4A10|TBC1D24	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000512795.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512795	ARFGEF2|ARX|COL18A1|COL4A1|CPT2|DCX|EMX2|FGFR3|FKRP|FKTN|FLNA|GPR56|LAMA2|LARGE|PAFAH1B1|PAX6|PEX7|POMGNT1|POMT1|POMT2|PQBP1|RAB3GAP1|RELN|SNAP29|SRPX2|TUBA1A|TUBA8|TUBB2B|WDR62	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000512796.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512796	ALDH7A1|ARFGEF2|ARHGEF9|ARX|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATRX|CACNA1A|CASK|CASR|CCDC88C|CDKL5|CHRNA2|CHRNA4|CHRNB2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COL18A1|COL4A1|CPT2|CSTB|CTSD|CUL4B|DCX|DEPDC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		5|DNAJC5|EFHC1|EMX2|EPM2A|FGD1|FGFR3|FKRP|FKTN|FLNA|FOXG1|GABRA1|GABRB3|GABRD|GABRG2|GPC3|GPR56|GRIA3|GRIN2A|HSD17B10|KCNA1|KCNJ1|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|LAMA2|LARGE|LBR|LGI1|MBD5|ME2|MECP2|MEF2C|MFSD8|NHLRC1|NIPBL|NOTCH3|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PCDH19|PEX7|PHF6|PIGV|PLA2G6|PLP1|PNKP|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRRT2|RAB39B|RAB3GAP1|RAI1|RELN|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SERPINI1|SETBP1|SLC25A22|SLC2A1|SLC4A10|SLC9A6|SMC1A|SMC3|SMS|SNAP29|SPTAN1|SRPX2|STXBP1|SYNGAP1|SYP|TBC1D24|TBX1|TCF4|TPP1|TREX1|TSC1|TSC2|TUBA1A|TUBA8|TUBB2B|UBE3A|VPS13A|VPS13B|WDR62|ZEB2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512797.2	Athena Diagnostics Inc — Massachusetts 22D0069726 512797	ATP2A2|ATP6V0A2|CCDC88C|CLCNKA|CLCNKB|KCNA1|KCNJ1|KCNJ10|KIAA1279|KMT2D|LBR|LGI1|NIPBL|PANK2|PIGV|PLA2G6|RAI1|SERPINI1|SETBP1|SMC3|SYNGAP1|TBX1|TSC1|TSC2|VPS13A|VPS13B	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515148.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515148	ARX	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515163.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515163	MEF2C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515170.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515170	SYNGAP1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515176.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515176	ARX	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515180.1	Athena	ARX	Deletion/duplication	Multiplex Ligation-dependent	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Inc — Massachusetts 22D0069726 515180		analysis	Probe Amplification (MLPA)			
GTR000515185.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515185	MEF2C	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000515191.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515191	MEF2C	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	No	Yes
GTR000515218.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515218	POMT2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515219.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515219	POMT1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515223.1	Athena Diagnostics Inc	FKTN	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	— Massachusetts 22D0069726 515223						
GTR000515235.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515235	COL6A1|COL6A2|COL6A3|FKRP|LAMA2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515238.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515238	COL6A1|COL6A2|COL6A3|FKRP|FKTN|LAMA2|POMGNT1|POMT1|POMT2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515246.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515246	CAPN3|CAV3|DMD|DYSF|FKRP|LMNA|MYOT|SGCA|SGCB|SGCD|SGCG	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515249.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515249	FKTN|POMGNT1|POMT1|POMT2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515254.1	Athena Diagnostics Inc —	FKRP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Massachusetts 22D0069726 515254						
GTR000515265.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515265	FKRP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515270.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515270	CAPN3|CAV3|DYSF|FKRP|LMNA|MYOT|SGCA|SGCB|SGCD|SGCG	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000515349.1	Athena Diagnostics Inc — Massachusetts 22D0069726 515349	POMGNT1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	Yes
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALD	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H6A1|ALDH7A1|ALDOA|ALDOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CACNA2D4|CASP8|CAT|CBS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1L|DPM1|DPM3|DSP|DTNBP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCTK|GM2A|GNAT1|GNAT2|GNE|GNPTAB|GNS|GOT1|GPD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		MC1R|MCCC1|MCCC2|MCEE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NEFH|NEUROD1|NFU1|NHS|NME1|NPHP1|NPHP3|NPHP4|NRL|NT5C3A|NUBPL|NYX|OAT|OCA2|OCRL|OGDH|OGG1|OPA1|OPA3|OPN1LW|OPN1MW|OPTN|OSTM1|OTC|OTX2|OXCT1|PAH|PANK2|PARK2|PARK7|PAX2|PAX6|PC|PCCA|PCCB|PCDH15|PCK2|PDE6A|PDE6B|PDE6C|PDE6G|PDHA1|PDHB|PDP1|PDSS1|PDSS2|PDZD7|PFKM|PGAM2|PGM1|PHB|PHKA1|PHKA2|PHKB|PHKG2|PHOX2A|PHYH|PINK1|PITPNM3|PITX2|PITX3|PLA2G2A|PLOD2|PLOD3|PMM2|PNKD|PNPLA2|POLG|POLG2|PPARG|PPARGC1B|PPI					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		B|PPOX|PPP2R1B|PRCD|PRKCG|PRODH|PROM1|PRPF31|PRPF6|PRPF8|PRPH2|PSAP|PSEN1|PTRF|PTS|PUS1|PYCR1|PYGL|PYGM|QDPR|RAF1|RARS2|RAX|RB1|RD3|RDH12|RDH5|REEP1|RET|RFT1|RGR|RGS9|RHO|RILP|RIMS1|RLBP1|RNASEL|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPL35A|RPS14|RRM2B|RS1|SAG|SARDH|SARS2|SCO2|SCP2|SDHAF1|SDHAF2|SDHB|SDHC|SECISBP2|SEMA4A|SEPT9|SERPINF1|SGSH|SHH|SIX6|SLC16A1|SLC22A4|SLC22A5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		TRPM1|TSFM|TSPAN12|TTC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513					
GTR000511381.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511381	ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ATP6V0A2|B4GALT1|COG1|COG7|COG8|DOLK|DPAGT1|DPM1|DPM3|GNE|MGAT2|MOGS|MPDU1|MPI|PMM2|RFT1|SLC35A1|SLC35C1|SRD5A3|TUSC3	Deletion/duplication analysis	Microarray	Not reported	Decline to answer	No
GTR000511382.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511382	ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ATP6V0A2|B4GALT1|COG1|COG7|COG8|DOLK|DPAGT1|DPM1|DPM3|GNE|MGAT2|MOGS|MPDU1|MPI|PMM2|RFT1|SLC35A1|SLC35C1|SRD5A3|TUSC3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	Decline to answer	No
GTR000511387.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511387	ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ATP6V0A2|B4GALT1|COG1|COG7|COG8|DOLK|DPAGT1|DPM1|DPM3|GNE|MGAT2|MOGS|MPDU1|MPI|PMM2|RFT1|SLC35A1|SLC35C1|SRD5A3|TUSC3	Sequence analysis of select exons	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511520.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511520	HSD17B10	Deletion/duplication analysis	Microarray	Not reported	Decline to answer	No
GTR000511521.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511521	HSD17B10	Sequence analysis of select exons	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000511522.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511522	HSD17B10	Sequence analysis of select exons	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000511523.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511523	HSD17B10	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511524.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511524	HSD17B10	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000511814.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511814	ARX	Deletion/duplication analysis|Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis|Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	Not reported	No
GTR000511815.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511815	ARX	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	Not reported	No
GTR000511816.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511816	ARX	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Not reported	Not reported	No
GTR000511817.1	Baylor Medical	ARX	Sequence analysis of the	Bi-directional Sanger	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511817		entire coding region	Sequence Analysis			
GTR000511818.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511818	ARX	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000511819.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511819	ARX	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512051.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512051	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512052.1	Baylor Medical Genetics	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratories Baylor College of Medicine Texas 45D0660090 512052						
GTR000512053.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512053	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No
GTR000512054.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512054	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512055.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512055	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512056.1	Baylor Medical Genetics Laboratories	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Baylor College of Medicine Texas 45D0660090 512056						
GTR000512057.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512057	MEF2C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512058.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512058	MEF2C	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512059.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512059	MEF2C	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000504097.1	Center for Genetics at Saint Francis Saint Francis	FGFR1|FGFR2|FGFR3	Sequence analysis of select exons|Targeted variant analysis	Bi-directional Sanger Sequence Analysis|SNP Detection	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Hospital Oklahoma 37D0474681 504097				(whole) blood|Product of conception (POC)		
GTR000504103.1	Center for Genetics at Saint Francis Saint Francis Hospital Oklahoma 37D0474681 504103	FGFR2|TWIST1	Sequence analysis of select exons|Targeted variant analysis	Bi-directional Sanger Sequence Analysis|SNP Detection	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000291094.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291094	FTSJ1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291100.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291100	TSPAN7	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291120.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291120	ACSL4	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000291122.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291122	ZNF41	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291134.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291134	KDM5C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291140.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291140	DLG3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291157.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291157	PAK3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291160.1	Center for Human Genetics, Inc — Massachusetts	ARHGEF6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	22D0650242 291160						
GTR000291163.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291163	AGTR2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291172.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291172	ATP6AP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000291505.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 291505	OPHN1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000319423.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 319423	GDI1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000321516.1	Center for Human Genetics,	SYNGAP1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Inc — Massachusetts 22D0650242 321516						
GTR000322960.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 322960	RAB39B	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	No
GTR000505960.1	Center for Human Genetics, Inc — Massachusetts 22D0650242 505960	ARX	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500883	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|BRD2|BTD|BUB1B|C12orf5	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		7|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDH2|IDS|IDUA|INPP5E|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASE					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000500936.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500936	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMAC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		R|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		0|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 503671	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|H					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|P					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		POX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510912.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 510912	ADSL|ALDH7A1|ARHGEF9|ARX|ATP6AP2|CACNB4|CDKL5|CHD2|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CSTB|CTSD|EFHC1|EPM2A|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GRIN2A|KANSL1|KCNJ10|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MECP2|MEF2C|MFSD	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fetal blood|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		8|NHLRC1|NRXN1|PCDH19|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRRT2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYN1|SYNGAP1|TBC1D24|TCF4|TPP1|TSC1|TSC2|UBE3A|ZEB2					
GTR000514881.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514881	AASS|ABAT|ABCD1|ABCD3|ABCD4|ACAD8|ACAD9|ACADM|ACADS|ACADVL|ACAT1|ACOX1|AGA|ALDH3A2|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMN|AMT|APTX|ARG1|ARSA|ASL|ASS1|ATP7A|ATP7B|AUH|B4GALT1|BCKDHA|BCKDHB|BTD|CACNA1H|CBS|CHAT|CISD2|CLN3|CLN5|CLN6|CLN8|COG1|COG4|COG5|COG6|COG7|COG8|COQ2|COQ9|CP|CPS1|CPT1A|CPT2|CTSA|CTSD|CTSF|CUBN|CYP27A1|DBH|DBT|DDC|DDOST|DHDDS|DHFR|DLAT|DLD|DNAJC5|DOLK|DPAGT1|DPM1|DPM2|DPM3|DPYD|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|ETFA|ETFB|ETFDH|ETHE1|FKRP|FKTN|FTCD|FUCA1|GAA|GALC|GAMT|GATM|GBA|GCDH|GCH1|GCSH|GIF|GK|GLB1|GLDC|GLUD1|GNE|GNPAT|GNPTAB|GRN|GUSB|HADH|HADHA|HADHB|HEXA|HEXB|HLCS|HMGCL|HMG	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CS2|HSD17B10|HYAL1|IDS|IDUA|IVD|KCTD7|L2HGDH|LARS2|LIPA|LMBRD1|MAGT1|MAN2B1|MANBA|MCCC1|MCCC2|MCEE|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|MUT|NAGA|NAGS|NDUFA1|NEU1|NPC1|NPC2|OTC|OXCT1|PAH|PCBD1|PCCA|PCCB|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHGDH|PHYH|PMM2|PNPO|POMGNT1|POMT1|POMT2|PPT1|PRPS1|PSAT1|PSPH|PTS|QDPR|RFT1|SLC16A2|SLC17A5|SLC19A2|SLC19A3|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A20|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A3|SLC6A4|SMPD1|SPR|SRD5A3|ST3GAL3|STT3A|STT3B|SUMF1|SUOX|TAT|TH|TIMM8A|TMEM165|TMLHE|TPH2|TPP1|TUBA1A					
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CR	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	514882	BN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000501305.1	Emory Genetics Laboratory Emory University School of Medicine Georgia	UPF3B	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	11D0683478 501305						
GTR000501308.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501308	UPF3B	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501310.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501310	IL1RAPL1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501311.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501311	IL1RAPL1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501313.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501313	PAK3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501314.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501314	PAK3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501315.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501315	ACSL4	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501316.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501316	ACSL4	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501334.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501334	GDI1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501335.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501335	ZNF81	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501336.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501336	GDI1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501337.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501337	ZNF81	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501339.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501339	POMT1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501340.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501340	POMT2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501341.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501341	POMT2	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501342.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501342	POMT1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501344.1	Emory Genetics	ARHGEF6	Sequence analysis of the	Bi-directional Sanger	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Emory University School of Medicine Georgia 11D0683478 501344		entire coding region	Sequence Analysis			
GTR000501345.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501345	ARX	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501346.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501346	TSPAN7	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501348.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501348	ARX	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501349.1	Emory Genetics Laboratory	ARHGEF6	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 501349						
GTR000501350.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501350	TSPAN7	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501351.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501351	AP1S2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501352.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501352	MBD5	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501353.1	Emory Genetics Laboratory Emory University	RAB39B	Mutation scanning of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	School of Medicine Georgia 11D0683478 501353						
GTR000501354.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501354	MBD5	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501355.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501355	AP1S2	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501356.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501356	RAB39B	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501445.1	Emory Genetics Laboratory Emory University School of	AGTR2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Medicine Georgia 11D0683478 501445						
GTR000501446.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501446	ZNF41	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501447.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501447	AGTR2	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501448.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501448	ZNF41	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501449.1	Emory Genetics Laboratory Emory University School of Medicine	ZDHHC15	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Georgia 11D0683478 501449						
GTR000501451.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501451	ZNF674	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501456.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501456	ZDHHC15	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501459.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501459	ZNF674	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501463.1	Emory Genetics Laboratory Emory University School of Medicine Georgia	BRWD3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	11D0683478 501463						
GTR000501468.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501468	GRIA3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501470.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501470	BRWD3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501471.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501471	GRIA3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501507.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478	CASK	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	501507						
GTR000501508.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501508	CASK	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501516.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501516	DLG3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501518.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501518	DLG3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501602.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501602	MAGT1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501605.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501605	MAGT1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501608.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501608	IGBP1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501610.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501610	OPHN1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501612.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501612	IGBP1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501613.1	Emory Genetics	OPHN1	Deletion/duplication	Comparative Genomic	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Emory University School of Medicine Georgia 11D0683478 501613		analysis	Hybridization			
GTR000501636.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501636	SOX3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501637.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501637	SRPX2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501638.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501638	SOX3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501639.1	Emory Genetics Laboratory	SRPX2	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 501639						
GTR000501640.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501640	ZDHHC9	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501641.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501641	ZNF711	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501642.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501642	ZDHHC9	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501643.1	Emory Genetics Laboratory Emory University	ZNF711	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	School of Medicine Georgia 11D0683478 501643						
GTR000501653.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501653	FTSJ1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501655.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501655	FTSJ1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501659.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501659	HSD17B10	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501660.1	Emory Genetics Laboratory Emory University School of	HSD17B10	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Medicine Georgia 11D0683478 501660						
GTR000501662.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501662	KDM5C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501664.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501664	KDM5C	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501665.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501665	KIAA2022	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501667.1	Emory Genetics Laboratory Emory University School of Medicine	KIAA2022	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Georgia 11D0683478 501667						
GTR000501708.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501708	ATP6AP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501711.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501711	ATP6AP2	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501732.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501732	MED12	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501733.1	Emory Genetics Laboratory Emory University School of Medicine Georgia	MED12	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	11D0683478 501733						
GTR000501734.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501734	FOXP1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501736.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501736	FOXP1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000501738.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501738	MEF2C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000501741.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478	MEF2C	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	501741						
GTR000502397.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502397	FKRP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502398.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502398	LARGE	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502450.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502450	FKRP	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502451.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502451	LARGE	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000502786.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502786	TUSC3	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502789.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502789	TUSC3	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502828.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502828	PHF8	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502830.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502830	PHF8	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502834.1	Emory Genetics	SYP	Sequence analysis of the	Bi-directional Sanger	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Emory University School of Medicine Georgia 11D0683478 502834		entire coding region	Sequence Analysis			
GTR000502835.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502835	SYP	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502836.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502836	UBE2A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502837.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502837	UBE2A	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502838.1	Emory Genetics Laboratory	HUWE1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 502838						
GTR000502839.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502839	HUWE1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502845.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502845	CUL4B	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000502846.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502846	CUL4B	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000503216.1	Emory Genetics Laboratory Emory University School of	COL6A1|COL6A2|COL6A3|FKRP|FKTN|ITGA7|LAMA2|LARGE|POMGNT1|POMT1|POMT2|SEPN1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Medicine Georgia 11D0683478 503216						
GTR000503227.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503227	COL6A1|COL6A2|COL6A3|FKRP|FKTN|ITGA7|LAMA2|LARGE|POMGNT1|POMT1|POMT2|SEPN1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000512325.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512325	ABCC9|ACTA2|ACTC1|ACTN2|ACVRL1|AKAP9|ANK2|ANKRD1|BAG3|BMPR2|BRAF|CACNA1C|CACNB2|CASQ2|CAV1|CAV3|CBS|COL3A1|COL5A1|COL5A2|CRYAB|CSRP3|DES|DMD|DSC2|DSG2|DSP|DTNA|EMD|ENG|FBN1|FBN2|FHL2|FKTN|FLNA|GATAD1|GLA|GPD1L|HCN4|HRAS|JPH2|JUP|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|KRAS|LAMA4|LAMP2|LDB3|LMNA|MAP2K1|MAP2K2|MED12|MYBPC3|MYH11|MYH6|MYH7|MYL2|MYL3|MYLK2|MYOZ2|MYPN|NEBL|NEXN|NKX2-5|NR2F2|NRAS|PKP2|PLN|PRKAG2|PTPN11|RAF1|RANGRF|RBM20|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SGCD|SKI|SLC2A10|SM	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		AD3|SNTA1|SOS1|TAZ|TCAP|TGFB2|TGFBR1|TGFBR2|TMEM43|TMPO|TNNC1|TNNI3|TNNT2|TPM1|TTN|TTR|VCL					
GTR000512331.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512331	ACTA2|CBS|COL3A1|COL5A1|COL5A2|FBN1|FBN2|FLNA|MED12|MYH11|SKI|SLC2A10|SMAD3|TGFB2|TGFBR1|TGFBR2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	No	No
GTR000512362.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512362	ABCA4|ADAM9|AIPL1|BEST1|C8orf37|CABP4|CACNA1F|CACNA2D4|CDHR1|CEP290|CERKL|CNGA3|CNGB3|CNNM4|CRX|GNAT2|GUCA1A|GUCA1B|GUCY2D|KCNV2|PAX6|PDE6C|PDE6H|PITPNM3|PROM1|PRPH2|RAX2|RBP4|RDH5|RGS9|RGS9BP|RIMS1|RPGR|RPGRIP1|SEMA4A|UNC119	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512371.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512371	B3GALTL|BCOR|BMP4|COL4A1|CYP1B1|FOXC1|FOXE3|FRAS1|FREM1|FREM2|GRIP1|HCCS|MFRP|NDP|OTX2|PAX6|PITX2|PITX3|SMOC1|SOX2|STRA6|VAX1|VSX2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512373.1	Emory Genetics Laboratory Emory University	ABCA4|ABHD12|ADAM9|AHI1|AIPL1|ALMS1|ARL13B|ARL6|B3GALTL|BBS1|BBS	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	School of Medicine Georgia 11D0683478 512373	10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCOR|BEST1|BMP4|C1QTNF5|C2orf71|C5orf42|C8orf37|CA4|CABP4|CACNA1F|CACNA2D4|CC2D2A|CDH23|CDH3|CDHR1|CEP290|CEP41|CERKL|CHM|CIB2|CLN3|CLN5|CLN6|CLN8|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|CNNM4|COL11A1|COL11A2|COL2A1|COL4A1|COL9A1|COL9A2|CRB1|CRX|CTSD|CYP1B1|DFNB31|DHDDS|EFEMP1|ELOVL4|EYS|FAM161A|FLVCR1|FOXC1|FOXE3|FRAS1|FREM1|FREM2|FSCN2|FZD4|GNAT1|GNAT2|GPR143|GPR179|GPR98|GRIP1|GRM6|GRN|GUCA1A|GUCA1B|GUCY2D|HARS|HCCS|IDH3B|IMPDH1|IMPG2|INVS|IQCB1|KCNJ13|KCNV2|KCTD7|KIF7|KLHL7|LCA5|LRAT|LRIT3|LRP5|LZTFL1|MAK|MERTK|MFN2|MFRP|MFSD8|MKKS|MKS1|MYO7A|MYOC|NDP|NPHP1|NPHP3|NPHP4|NR2E3|NRL|NYX|OCA2|OFD1|OPA1|OPA3|OTX2|PAX6|PCDH15|PDE6A|PDE6B|PDE6C|PDE6G|PDE6H|PDZD7|PEX7|PHYH|PITPNM3|PITX2|PITX3|PLA2G5|PPT1|PRCD|PROM1|PRPF3|PRPF31|PRPF6|PRPF8|PRPH2|RAX2|RBP3|RBP4|RD3|RDH12|RDH5|RGR|RGS9|RGS9BP|RHO|R		parallel sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		IMS1|RLBP1|ROM1|RP1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPGRIP1L|RS1|SAG|SDCCAG8|SEMA4A|SLC24A1|SLC45A2|SMOC1|SNRNP200|SOX2|SPATA7|STRA6|TCTN1|TCTN2|TCTN3|TIMM8A|TIMP3|TMEM126A|TMEM216|TMEM237|TMEM67|TOPORS|TPP1|TRIM32|TRPM1|TSPAN12|TTC21B|TTC8|TULP1|TYR|TYRP1|UNC119|USH1C|USH1G|USH2A|VAX1|VCAN|VSX2|WDPCP|WT1|ZNF423|ZNF513					
GTR000512377.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512377	ABCA4|BEST1|CDH3|CNGB3|EFEMP1|ELOVL4|FSCN2|GUCA1B|PROM1|PRPH2|RBP4|RDH12|RPGR|RPGRIP1|TIMP3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512415.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512415	ACVR2B|AHI1|AIPL1|ARL13B|ARL6|ATXN10|B9D1|B9D2|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|C2orf71|C5orf42|CC2D2A|CCDC28B|CCDC39|CCDC40|CDH23|CEP164|CEP290|CEP41|CFTR|CLRN1|CRB1|CRELD1|CRX|DFNB31|DNAAF1|DNAAF2|DNAAF3|DNAH11|DNAH5|DNAI1|DNAI2|DNAL1|DYNC2H1|EVC|EVC2|FOXH1|GDF1|GLIS2|GPR98|GUCY2D	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|HYLS1|IFT43|IFT80|IMPDH1|INVS|IQCB1|KCNJ13|KIF7|LCA5|LEFTY2|LRAT|MKKS|MKS1|MYO7A|NEK1|NEK8|NKX2-5|NME8|NODAL|NPHP1|NPHP3|NPHP4|OFD1|PCDH15|PKD2|PKHD1|RD3|RDH12|RPE65|RPGR|RPGRIP1|RPGRIP1L|RSPH4A|RSPH9|SCNN1A|SCNN1B|SCNN1G|SDCCAG8|SPATA7|TCTN1|TCTN2|TMEM138|TMEM216|TMEM231|TMEM237|TMEM67|TOPORS|TRIM32|TSC1|TSC2|TTC21B|TTC8|TULP1|UMOD|USH1C|USH1G|USH2A|VHL|WDPCP|WDR19|WDR35|XPNPEP3|ZIC3|ZNF423					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512416.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512416	ABAT|ADSL|ALDH7A1|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|BCKDK|CACNA1A|CACNB4|CASK|CASR|CDKL5|CENPJ|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DNAJC5|EFHC1|EMX2|EPM2A|FLNA|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GPR56|GPR98|GRIN2A|HCN1|HCN4|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MTHFR|NDE1|NDUFA1|NHLRC1|NRXN1|OPHN1|PAFAH1B1|PCDH19|PHF6|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRICKLE2|PRRT2|RELN|SCARB2|SCN1A|SCN1B|SCN2A|SCN3A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TPP1|TSC1|TSC2|TSEN54|UBE3A|WDR62|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512417.1	Emory Genetics Laboratory Emory University School of Medicine Georgia	ADAM17|AICDA|ATG16L1|BTK|C1orf106|CD40LG|COL7A1|CYBA|CYBB|DCLRE1C|DOCK8|ERAP2|FERMT1|FOXP3|FUT2|G6PC3|GUCY2C|HPS1|HPS4|HPS6|I	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	11D0683478 512417	COS|IL10|IL10RA|IL18RAP|IL23R|IL2RA|IRGM|ITGAM|LRBA|MEFV|MVK|NCF2|NCF4|NOD2|PIK3R1|PLCG2|PTEN|PTPN22|RAC2|RB1|RET|SH2D1A|SLC37A4|STAT1|STXBP2|TTC37|WAS|XIAP					
GTR000512422.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512422	ACTA1|AMPD1|AMPD3|ANO5|CAPN3|CAV3|COL6A1|COL6A2|COL6A3|DES|DMD|DYSF|EMD|FKRP|FKTN|GAA|GNE|ISPD|ITGA7|LAMA2|LARGE|LMNA|MYOT|NEB|PLEC|PMM2|POMGNT1|POMT1|POMT2|PYGM|RYR1|RYR2|SEPN1|SGCA|SGCB|SGCD|SGCE|SGCG|SIL1|TCAP|TNNI2|TNNT1|TPM2|TPM3|TRIM32|TTN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512423.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512423	ATRX|BLM|BTK|CREBBP|CUL7|DHCR7|EP300|ERCC6|ERCC8|FGD1|GH1|GHR|GHRHR|GLI2|HESX1|IGF1|IGF1R|INSR|KDM6A|KMT2D|KRAS|LHX3|NBN|NIPBL|PITX2|POU1F1|PROP1|PTPN11|RAF1|ROR2|RPS6KA3|SHOX|SHOX2|SMARCAL1|SMC1A|SMC3|SOS1|SOX2|SOX3|SRCAP|STAT5B|TBCE|THRB|TRIM37|WRN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512586.1	Emory Genetics Laboratory Emory University School of	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK16|CDKL5|CLIC2|CNKSR2|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Medicine Georgia 11D0683478 512586	CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|HCFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711				
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512588.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512588	ACTB|ACTG1|AHI1|ARFGEF2|ARX|CASK|CC2D2A|CEP290|CEP41|CHMP1A|DCX|EOMES|EXOSC3|FKRP|FKTN|FLNA|GPR56|KIAA1279|KIF7|LAMC3|LARGE|MKS1|NPHP1|OPHN1|PAFAH1B1|POMGNT1|POMT1|POMT2|PQBP1|RAB3GAP1|RAB3GAP2|RARS2|RELN|RPGRIP1L|RTTN|SRPX2|TMEM138|TMEM216|TMEM237|TMEM67|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|VRK1|WDR62	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512591.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512591	ALG1|ALG11|ALG12|ALG13|ALG14|ALG3|ALG6|ALG8|ALG9|ATP6V0A2|B3GALTL|B3GAT3|B4GALT1|B4GALT7|CHST14|CHST3|CHST6|CHSY1|COG1|COG4|COG5|COG6|COG7|COG8|DDOST|DHDDS|DOLK|DPAGT1|DPM1|DPM3|EXT1|EXT2|FKRP|FKTN|GALNT3|GFPT1|GNE|LARGE|LFNG|MAN1B1|MGAT2|MOGS|MPDU1|MPI|NGLY1|PGM1|PIGA|PIGL|PIGM|PIGO|PIGV|PMM2|POMGNT1|POMT1|POMT2|RFT1|RPN2|SEC23B|SLC35A1|SLC35C1|SLC35D1|SRD5A3|ST3GAL3|ST3GAL5|TMEM165|TUSC3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512592.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512592	CHKB|COL6A1|COL6A2|COL6A3|DAG1|DPM2|DPM3|FKRP|FKTN|ISPD|ITGA7|LAMA2|LARGE|LMNA|POMGNT2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512595.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512595	ACP5|ANKH|ANO5|B3GALT6|BMPR1B|CANT1|CDKN1C|CHST14|CHST3|COL10A1|COL11A1|COL11A2|COL1A1|COL2A1|COL9A1|COL9A2|COL9A3|COMP|CTSK|CUL7|DDR2|DHCR24|DYM|DYNC2H1|EIF2AK3|EVC|EVC2|EXT1|EXT2|FAM20C|FBN1|FGFR1|FGFR2|FGFR3|FLNA|FLNB|GDF5|GLI3|GPC6|HSPG2|ICK|IFT122|IFT140|IFT80|IHH|KIF22|LIFR|MATN3|MMP13|MMP9|NEK1|NKX3-2|NPR2|OBSL1|PAPSS2|PCNT|PRKAR1A|PTH1R|PTPN11|ROR2|RUNX2|SH3PXD2B|SHOX|SLC25A12|SLC26A2|SLC35D1|SLC39A13|SMARCAL1|SOX9|SULF1|TBCE|TCTN3|TRIP11|TRPS1|TRPV4|WDR35|WNT5A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512597.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512597	ACTA1|AMPD1|AMPD3|ANO5|BAG3|BIN1|BSCL2|CAPN3|CAV3|CFL2|CHAT|CHRNA1|CHRNB1|CHRND|CHRNE|CHRNG|COL6A1|COL6A2|COL6A3|COLQ|CRYAB|DAG1|DES|DMD|DNM2|DOK7|DYSF|EMD|FHL1|FKRP|FKTN|FLNC|GAA|GLE1|GNE|IGHMBP2|ISPD|ITGA7|LAMA2|LARGE|LDB3|LMNA|MTM1|MTMR14|MUSK|MYH2|MYH7|MYOT|NEB|PABPN1|PLEC|PLEKHG5|PMM2|POMGNT1|POMT1|POMT2|PTRF|PYGM|RAPSN|RYR1|RYR2|SCN4A|SEPN1|SGCA|SGCB|SGCD|SGCE|SGCG|SIL1|SYNE1|SYNE2|TCAP|TNNI2|TNNT1|TPM2|TPM3|TRIM32|TTN|VRK1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512601.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512601	ABHD12|ABHD5|ACTG1|ARSB|ATP6V1B1|BSND|BTD|CCDC50|CDH23|CEACAM16|CISD2|CLDN14|CLRN1|COCH|COL11A2|COL4A5|CRYM|DFNA5|DFNB31|DFNB59|DIAPH1|DNMT1|DSPP|ESRRB|EYA1|EYA4|FGFR3|FOXC1|FOXI1|GIPC3|GJB2|GJB3|GJB6|GPR98|GPSM2|GRHL2|GRXCR1|HARS2|HGF|HSD17B4|ILDR1|KCNE1|KCNJ10|KCNQ1|KCNQ4|LHFPL5|LOXHD1|LRTOMT|MARVELD2|MASP1|MSRB3|MYH14|MYH9|MYO15A|MYO1A|MY	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		O3A|MYO6|MYO7A|OTOA|OTOF|PCDH15|PDZD7|PITX2|POU3F4|PRPS1|RPS6KA3|SALL1|SALL4|SERPINB6|SIX1|SLC17A8|SLC26A4|SLC26A5|SLC29A3|SMPX|TECTA|TIMM8A|TJP2|TMC1|TMIE|TMPRSS3|TPRN|TRIOBP|USH1C|USH1G|USH2A|WFS1					
GTR000512602.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512602	DOCK8|SPINK5|STAT3|TYK2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512604.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512604	ARHGAP31|BMP2|BMPR1B|CC2D2A|CDH3|CEP290|CHSY1|ESCO2|FBLN1|FBXW4|FGF10|FGFR2|FGFR3|FMN1|GDF5|GLI3|GNAS|GREM1|HDAC4|HOXD13|IHH|KIF7|LMBR1|LRP4|MGP|MKS1|MYCN|NIPBL|NOG|PIGV|PITX1|PTHLH|RECQL4|ROR2|RPGRIP1L|SALL1|SALL4|SHH|SOX9|TBX15|TBX3|TBX5|THPO|TP63|WNT3|WNT7A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512605.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512605	ANO5|CAPN3|CAV3|DAG1|DES|DYSF|FKRP|FKTN|FLNC|LMNA|MYOT|PLEC|POMGNT1|POMT1|POMT2|SGCA|SGCB|SGCD|SGCG|TCAP|TRIM32|TTN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512606.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512606	CUL4B|EZH2|GLI3|GPC3|MED12|NFIX|NSD1|PHF6|PTCH1|PTEN|UPF3B	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512608.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512608	ACTB|ACTG1|ADSL|AHI1|ALDH7A1|ARFGEF2|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|ATR|ATRX|BCKDK|CACNB4|CASC5|CASK|CC2D2A|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP290|CEP41|CEP63|CHMP1A|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DHCR7|DISP1|DNAJC5|EFHC1|EHMT1|EOMES|EPM2A|EXOSC3|FGF8|FKRP|FKTN|FLNA|FOLR1|FOXG1|FOXH1|GABRA1|GABRG2|GAMT|GATM|GLI2|GOSR2|GPR56|GPR98|GRIN2A|GRIN2B|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCN	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		T1|KCTD7|KIAA1279|KIF7|LAMC3|LARGE|LARP7|LGI1|LIAS|MAGI2|MAPK10|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MKS1|MYCN|NDE1|NHLRC1|NIN|NODAL|NPHP1|NRXN1|OPHN1|ORC4|ORC6|PAFAH1B1|PCDH19|PCNT|PLCB1|PNKP|PNPO|POC1A|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRRT2|PTCH1|RAB18|RAB3GAP1|RAB3GAP2|RARS2|RBBP8|RELN|RPGRIP1L|RTTN|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TGIF1|TMEM138|TMEM216|TMEM237|TMEM67|TPP1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|UBE3A|VLDLR|VRK1|WDR62|ZEB2|ZIC2|ZNF335					
GTR000512610.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512610	ABCA3|ACVRL1|AP3B1|ASCL1|BDNF|BLOC1S3|BLOC1S6|BMPR2|CCDC39|CCDC40|CFTR|CSF2RA|DNAAF1|DNAAF2|DNAH11|DNAH5|DNAI1|DNAI2|DNAL1|DOCK8|DTNBP1|EDN3|EFEMP2|ELMOD2|ELN|ENG|FBLN5|FLCN|GDNF|HPS1|HPS3|HPS4|HPS5|HPS6|LTBP4|MUC5B|NME8|PHO	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		X2B|RET|RSPH4A|RSPH9|SCNN1A|SCNN1B|SCNN1G|SERPINA1|SFTPA1|SFTPA2|SFTPB|SFTPC|SFTPD|SMAD9|STAT3|TERT|TSC1|TSC2					
GTR000512649.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512649	ACP5|ALPL|ANKH|ANO5|ARHGAP31|ATP6V0A2|B3GALT6|B4GALT7|BMP2|BMPR1B|CA2|CANT1|CASR|CC2D2A|CDH3|CDKN1C|CEP290|CHST14|CHST3|CHSY1|CLCN5|CLCN7|COL10A1|COL11A1|COL11A2|COL1A1|COL1A2|COL2A1|COL9A1|COL9A2|COL9A3|COMP|CRTAP|CTSK|CUL7|DDR2|DHCR24|DLX3|DMP1|DYM|DYNC2H1|EIF2AK3|ENPP1|ESCO2|EVC|EVC2|EXT1|EXT2|FAM20C|FBLN1|FBN1|FBXW4|FERMT3|FGF10|FGF23|FGFR1|FGFR2|FGFR3|FKBP10|FLNA|FLNB|FMN1|GALNT3|GDF5|GLI3|GNAS|GORAB|GPC6|GREM1|HDAC4|HOXD13|HPGD|HSPG2|ICK|IFITM5|IFT122|IFT140|IFT80|IHH|KIF22|KIF7|LEMD3|LEPRE1|LIFR|LMBR1|LMNA|LRP4|LRP5|MAFB|MATN3|MGP|MKS1|MMP13|MMP2|MMP9|MYCN|NEK1|NIPBL|NKX3-2|NOG|NOTCH2|NPR2|OBSL1|OSTM1|PAPSS2|PCNT|PHEX|PIGV|PITX1|PLOD2|PPIB|PRKAR1A|PTH1R|PTHLH|PTPN11|PYCR1|R	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ASGRP2|RECQL4|ROR2|RPGRIP1L|RUNX2|SALL1|SALL4|SERPINH1|SH3PXD2B|SHH|SHOX|SLC25A12|SLC26A2|SLC34A3|SLC35D1|SLC39A13|SMARCAL1|SOST|SOX9|SP7|SULF1|TBCE|TBX15|TBX3|TBX5|TBXAS1|TCIRG1|TCTN3|TGFB1|THPO|TMEM216|TMEM67|TNFRSF11A|TNFRSF11B|TNFSF11|TP63|TREM2|TRIP11|TRPS1|TRPV4|TYROBP|WDR35|WISP3|WNT3|WNT5A|WNT7A|ZMPSTE24					
GTR000505642.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 505642	ACTA2|CBS|COL3A1|COL5A1|COL5A2|FBN1|FBN2|FLNA|MED12|MYH11|MYLK|SKI|SLC2A10|SMAD3|TGFB2|TGFBR1|TGFBR2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000505662.12	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 505662	ANO5|CAPN3|CAV3|DAG1|DES|DMD|DNAJB6|DPM3|DYSF|EMD|FHL1|FKRP|FKTN|LMNA|MYOT|POMGNT1|POMT1|POMT2|PTRF|SGCA|SGCB|SGCD|SGCG|SYNE1|SYNE2|TCAP|TMEM43|TRIM32|TTN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000506412.12	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189	ABCA4|ADAM9|AHI1|AIPL1|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCOR|BEST1|C2orf71|CA4|CABP4|CACNA1F|CACNA2D4|CC2D2A|CDH23|C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	506412	DH3|CEP290|CERKL|CHM|CLN3|CNGA1|CNGB1|CNGB3|CNNM4|COL11A1|COL2A1|COL9A1|CRB1|CRX|CYP1B1|CYP4V2|DFNB31|EFEMP1|ELOVL4|EYS|FAM161A|FOXC1|FSCN2|GNAT1|GRM6|GUCA1A|GUCA1B|GUCY2D|HMCN1|IDH3B|IMPDH1|IMPG2|INVS|IQCB1|JAG1|KCNJ13|KCNV2|KLHL7|LCA5|LRAT|MERTK|MFRP|MKKS|MTTP|MYOC|NDP|NPHP1|NPHP3|NPHP4|NR2E3|NRL|OAT|OPA1|OPA3|OPTN|OTX2|PANK2|PAX2|PAX6|PCDH15|PDE6A|PDE6B|PDE6C|PDE6G|PEX1|PEX2|PEX7|PHYH|PITPNM3|PITX2|PRCD|PROM1|PRPF3|PRPF31|PRPF8|PRPH2|RAX2|RB1|RBP3|RBP4|RD3|RDH12|RGR|RGS9|RGS9BP|RHO|RIMS1|RLBP1|ROM1|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPGRIP1L|SAG|SEMA4A|SNRNP200|SOX2|SPATA7|TIMM8A|TIMP3|TMEM126A|TOPORS|TREX1|TRIM32|TRPM1|TTPA|TULP1|UNC119|USH1C|VCAN|VSX2|WFS1|ZNF513					
GTR000506437.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California	ABCC9|ACTC1|ACTN2|AKAP9|ANK2|ANKRD1|BAG3|CACNA1C|CACNA2D1|CACNB2|CALR3|CASQ2|CAV3|COX15|CRYAB|CSRP3|DES|DMD|DOLK|DSC2|DS	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	05D2043189 506437	G2|DSP|DTNA|EMD|EYA4|FKTN|FLNA|FXN|GAA|GATA4|GATAD1|GJA5|GLA|GPD1L|HCN4|ILK|JAG1|JPH2|JUP|KCNA5|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|LAMA4|LAMP2|LDB3|LMNA|MRPL3|MURC|MYBPC3|MYH6|MYH7|MYL2|MYL3|MYLK2|MYOM1|MYOZ2|MYPN|NDUFAF1|NEBL|NEXN|NKX2-5|NPPA|PDLIM3|PKP2|PLN|PRKAG2|PSEN2|PTPN11|RAF1|RBM20|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SDHA|SGCD|SNTA1|SYNE1|TAZ|TBX1|TBX5|TCAP|TGFB3|TMEM43|TMPO|TNNC1|TNNI3|TNNT2|TPM1|TTN|TTR|TXNRD2|VCL					
GTR000507860.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 507860	AARS|ATL1|DNM2|DNMT1|DYNC1H1|EGR2|FGD4|GARS|GDAP1|GJB1|GLA|HSPB1|HSPB8|KIF1B|LITAF|LMNA|LRSAM1|MED25|MFN2|MPZ|MTMR2|NDRG1|NEFL|PMP22|PRX|RAB7A|SBF2|SCN9A|SH3TC2|SPTLC2|TRPV4|TTR|YARS	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000508339.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 508339	APC|ATM|ATR|AXIN2|BAP1|BARD1|BLM|BMPR1A|BRCA1|BRCA2|BRIP1|CDH1|CDK4|CDKN2A|CHEK2|CTNNB1|EPCAM|FANCC|HOXB13|MLH1|MRE11A|MSH2|MSH6|MUTYH|NBN|PALB2|PALLD|PMS2|PTEN|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		RAD50|RAD51|RAD51C|RAD51D|SMAD4|STK11|TP53|VHL|XRCC2|XRCC3					
GTR000508744.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 508744	CASK|OPHN1|RARS2|SEPSECS|TSEN2|TSEN34|TSEN54|VRK1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6					
GTR000509367.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509367	CASK|OPHN1|RARS2|SEPSECS|TSEN2|TSEN34|TSEN54|VRK1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509368.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509368	ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|ATP6V0A2|B3GALTL|B4GALT1|COG1|COG4|COG5|COG6|COG7|COG8|DDOST|DHDDS|DOLK|DPAGT1|DPM1|DPM3|GNE|MAGT1|MGAT2|MOGS|MPDU1|MPI|PMM2|RFT1|RPN2|SEC23B|SLC35A1|SLC35C1|SRD5A3|TUSC3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509399	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|V					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000509426.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509426	ACVR1|BEST1|CA4|CANT1|COL18A1|CYP1B1|FOXC1|ISPD|LMX1B|LOXL1|LTBP2|MTHFR|MYOC|NTF4|OPA1|OPTN|PAX6|PITX3|POMT1|RPS19|RRM2B|SBF2|SLC4A4|TTR|WDR36	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509427.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509427	AIP|APC|ATM|ATR|AXIN2|BAP1|BARD1|BLM|BMPR1A|BRCA1|BRCA2|BRIP1|BUB1B|CDH1|CDK4|CDKN1B|CDKN2A|CHEK2|CTNNB1|CYLD|DDB2|DICER1|EGFR|EGLN1|EPCAM|ERCC2|ERCC3|ERCC4|ERCC5|EXO1|EXT1|EXT2|FANCA|FANCB|FANCC|FANCD2|FANCE|FANCF|FANCG|FANCI|FANCL|FANCM|FH|FLCN|GALNT12|GPC3|HOXB13|HRAS|KIF1B|KIT|MAX|MC1R|MEN1|MET|MITF|MLH1|MPL|MRE11A|MSH2|MSH3|MSH6|MUTYH|NBN|NF1|NF2|PALB2|PDGFRA|PICALM|PMS1|PMS2|POLD1|PRKAR1A|PRKDC|PRSS1|PTCH1|PTEN|PTPN11|RAD50|RAD51|RAD51C|RAD51D|RB1|RBBP8|RBM15|RECQL4|RET|ROBO2|SBDS|SDHA|SDHAF2|SDHB|SDHC|SDHD|SLX4|SMAD4|SMARCB1|STK11|SUFU|TERT|TMEM127|TP53|TSC1|TSC2|TSHR|TYR|VHL|WR	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		N|WT1|XPA|XPC|XRCC2|XRCC3					
GTR000509430.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509430	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|LARGE|PAFAH1B1|POMGNT1|POMT1|POMT2|RELN|TUBA1A|VLDLR	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509431.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509431	ABAT|ACOX1|ALDH3A2|ARSA|ASPA|CSF1R|DARS2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|FAM126A|GFAP|GJC2|HEPACAM|HSPD1|HTRA1|LMNB1|MLC1|NOTCH3|PLP1|PSAP|PTEN|SCP2|SLC25A12	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509432.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509432	ACAD9|ADCK3|APTX|ATP5E|ATPAF2|BCS1L|COQ2|COQ9|COX10|COX15|COX6B1|DARS2|DGUOK|DLAT|DLD|DNM1L|ETFA|ETFB|ETFDH|ETHE1|FBP1|FH|FOXRED1|G6PC|GFM1|GYS2|ISCU|LRPPRC|MRPS16|MRPS22|NDUFA11|NDUFAF1|NDUFAF3|NDUFAF4|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|PC|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|POLG|PUS1|RRM2B|SCO2|SLC25A3|SLC25A4|SUCLA2|SUCLG1|TAZ|TK2|TMEM70|TRMU|TSFM|TUFM|TYMP|UQCRB|YARS2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509436.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509436	ABCC8|ACY1|ADAMTSL2|ADSL|AGA|ALDH4A1|ALDH5A1|ALDH7A1|AMT|ANTXR2|ARG1|ARSA|ARSB|ASAH1|ASPA|ATP13A2|BTD|CLN3|CLN5|CLN6|CLN8|COL11A2|COL2A1|CTNS|CTSA|CTSC|CTSD|CTSK|DHCR7|DNAJC5|DPYD|DYM|ETFA|ETFB|ETFDH|FH|FOLR1|FUCA1|GAA|GALC|GALNS|GAMT|GBA|GCDH|GCSH|GLA|GLB1|GLDC|GM2A|GNE|GNPTAB|GNPTG|GNS|GPC3|GUSB|HEXA|HEXB|HGSNAT|HPD|HRAS|IDS|IDUA|L2HGDH|LAMA2|LAMP2|LIPA|LMBRD1|MAN2B1|MANBA|MCOLN1|MFSD8|MOCS1|MOCS2|NAGA|NAGLU|NEU1|NPC1|NPC2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX26|PEX3|PEX5|PEX6|PGK1|PHYH|PPT1|PRODH|PSAP|QDPR|RAI1|SGSH|SLC17A5|SLC25A15|SLC46A1|SMPD1|SUMF1|SUOX|TCF4|TPP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509441.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509441	ABCA4|C2|C3|CFB|CFH|CFI|CNGB3|CST3|CX3CR1|EFEMP1|ELOVL4|ERCC6|FBLN5|HMCN1|HTRA1|PRPH2|RAX2|RLBP1|RPGR|TLR4	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509442.11	Fulgent Clinical Diagnostics Lab	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation		Not reported	Not reported

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509442	ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|	of the entire coding region	(NGS)/Massively parallel sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|R					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509443.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189	ALDH7A1|ARX|ATP1A2|CACNA1A|CDKL5|FOLR1|FOXG1|GAMT|KCNQ2|MECP2|PCDH19|PHGDH|PNPO|POLG|PPT1|SCN1A|SLC2A1|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	509443						
GTR000509444.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509444	ACAD9|ACADL|ACADM|ACADVL|AGL|C10orf2|CPT1B|CPT2|GAA|GYS1|HADHA|HADHB|OPA1|OPA3|PFKM|PGAM2|PGM1|PHKA1|POLG|POLG2|RRM2B|SUCLA2|TK2|TYMP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509446.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509446	C10orf2|DGUOK|MPV17|POLG|POLG2|RRM2B|SLC25A4|SUCLA2|SUCLG1|TK2|TYMP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509449.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509449	AKT3|BUB1B|CASK|CDK5RAP2|CENPJ|CEP135|CEP152|CEP63|DNM1L|EFTUD2|IER3IP1|KIF11|LIG4|MCPH1|MED17|MRE11A|MSMO1|NDE1|NHEJ1|NR2E1|PAFAH1B1|PCNT|PNKP|POMT1|PQBP1|RARS2|SLC25A19|STIL|TSEN2|TSEN34|TSEN54|TUBB2B|TUBGCP6|VRK1|WDR62	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509453.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509453	CLN3|CLN5|CLN6|CLN8|CTSD|DNAJC5|KCTD7|MFSD8|PPT1|TPP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509454.8	Fulgent Clinical	ACTA1|AMPD1|AMPD3|AN	Deletion/duplication	Next-Generation	Buccal	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Lab Fulgent Diagnostics California 05D2043189 509454	O5|CAPN3|CAV3|COL6A1|COL6A2|COL6A3|DES|DMD|DYSF|EMD|FKRP|FKTN|ITGA7|LAMA2|LARGE|LMNA|MYOT|NEB|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PLEC|PMM2|POMGNT1|POMT1|POMT2|RYR1|RYR2|SEPN1|SGCA|SGCB|SGCD|SGCE|SGCG|SIL1|TCAP|TNNI2|TNNT1|TPM2|TPM3|TRIM32|TTN	analysis|Sequence analysis of the entire coding region	(NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	swab|Isolated DNA|Peripheral (whole) blood|Saliva	answer	
GTR000509461.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509461	ATR|CDC6|CDT1|CENPJ|CEP152|CEP63|ORC1|ORC4|ORC6|PCNT|RBBP8|RNU4ATAC	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509462.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509462	ARX|ATRX|CAV1|CDKL5|CNTNAP2|FOXG1|MECP2|MED17|MEF2C|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|TRAPPC9|UBE3A|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509491.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509491	GPR56|KIAA1279|SRPX2|TUBA1A|TUBA8|TUBB2B|TUBB3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509492.7	Fulgent Clinical	HESX1|OTX2|PAX6|PROP	Deletion/duplication	Next-Generation	Buccal	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Lab Fulgent Diagnostics California 05D2043189 509492	1|SOX2	analysis|Sequence analysis of the entire coding region	(NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	swab|Isolated DNA|Peripheral (whole) blood|Saliva	answer	
GTR000510709.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510709	ACTB|ACTG1|ATP2B2|ATP6V1B1|BCS1L|BSND|CATSPER2|CCDC50|CDH23|CLDN14|CLRN1|COCH|COL11A2|COL9A3|CRYM|DFNA5|DFNB31|DFNB59|DIAPH1|DSPP|EDN3|EDNRB|ERCC2|ERCC3|ESPN|ESRRB|EYA1|EYA4|FGF3|FOXI1|GATA3|GIPC3|GJA1|GJB2|GJB3|GJB6|GPR98|GPSM2|GRHL2|GRXCR1|GSTP1|HGF|ILDR1|JAG1|KCNE1|KCNJ10|KCNQ1|KCNQ4|LHFPL5|LHX3|LOXHD1|LRTOMT|MARVELD2|MIR96|MITF|MSRB3|MTAP|MYH14|MYH9|MYO15A|MYO1A|MYO1C|MYO1F|MYO3A|MYO6|MYO7A|OTOA|OTOF|PAX3|PCDH15|PDZD7|PMP22|POU3F4|POU4F3|PRPS1|PTPRQ|RDX|SERPINB6|SIX1|SLC17A8|SLC26A4|SLC26A5|SLC4A11|SMPX|SNAI2|SOX10|SPINK5|TBL1X|TCF21|TECTA|TIMM8A|TJP2|TMC1|TMIE|TMPRSS3|TMPRSS5|TPRN|TRIOBP|TRMU|USH1C|USH1G|USH2A|WFS1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510870.7	Fulgent Clinical	DHFR|FOLR1|FPGS|MTHF	Deletion/duplication	Next-Generation	Buccal	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Lab Fulgent Diagnostics California 05D2043189 510870	D1|MTHFD1L|MTHFS|PTS|SHMT1|SLC19A1	analysis|Sequence analysis of the entire coding region	(NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	swab|Isolated DNA|Peripheral (whole) blood|Saliva	answer	
GTR000510871.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510871	AKAP9|ANK2|CACNA1C|CACNB2|CALR3|CASQ2|CAV3|CSRP3|DES|DSC2|DSG2|DSP|DTNA|EYA4|FBN1|FBN2|FKTN|GJA5|GPD1L|JPH2|JUP|KCNA5|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNQ1|LAMP2|LDB3|LMNA|LRP6|MYBPC3|MYH6|MYH7|MYL2|MYL3|MYLK2|MYOZ2|NEXN|NPPA|PKP2|PLN|PRKAG2|PSEN1|PSEN2|RBM20|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SGCD|SLC25A4|SNTA1|TAZ|TCAP|TGFB3|TGFBR2|TMEM43|TMPO|TNNC1|TNNI3|TNNT2|TPM1|TTN|VCL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510909.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510909	ABCD1|ACOX1|ATL1|BSCL2|CYP7B1|FA2H|KIAA0196|KIF1A|KIF5A|L1CAM|NIPA1|OPA3|PLP1|REEP1|RTN2|SLC16A2|SLC33A1|SPAST|SPG11|SPG20|SPG21|SPG7|ZFYVE26|ZFYVE27	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	California 05D2043189 510915	ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACSM3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKDHA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|CLYBL|CNR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI		parallel sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYCTK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|M					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		TRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISB					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510916.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510916	ABCD1|ACSL4|AFF2|AGTR2|AIFM1|AP1S2|ARHGEF6|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|EBP|FAAH2|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|GSPT2|HCCS|HDAC8|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|L1CAM|LAMP2|MAGT1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NDUFA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RAB40AL|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SRPX2|SYN1|SYP|TAF1|TIMM8A|TSPAN7|UBE2A|UPF3B|USP9X|WDR13|ZCCHC12|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000514645.1	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 514645	ACAD9|ACADL|ACADM|ACADS|ACADVL|CPT1A|CPT1B|CPT2|ETFA|ETFB|ETFDH|GLUD1|HADH|HADHA|HADHB|HMGCL|HMGCS2|HSD17B10|LPIN1|SLC22A5|SLC25A20|TAZ	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cell culture|Isolated DNA|Paraffin block|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000310785.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 310785	OPHN1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000324198.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 324198	MEF2C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000324273.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 324273	CASK	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000325792.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 325792	WDR62	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000332148.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 332148	GRIN2A	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500125.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500125	CASK	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500138.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500138	MEF2C	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500153.5	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500153	ARHGEF9|ARX|CDKL5|EFHC1|GRIN2A|KCNQ2|PCDH19|PLCB1|PNKP|POLG|SCN1A|SCN2A|SCN8A|SLC25A22|SLC2A1|SPTAN1|ST3GAL3|ST3GAL5|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000500154.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500154	ALG6|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|C12orf57|CA8|CC2D1A|CNTNAP2|CRBN|D2HGDH|DDHD2|ERLIN2|GRIK2|IDH2|KCNJ10|L2HGDH|LINS|LRP2|MAN1B1|MED23|NRXN1|NSUN2|PCNT|PRSS12|SLC25A1|ST3GAL3|STXBP1|TECR|TRAPPC9|TUSC3|VLDLR|VPS13B|ZC3H14|ZNF526	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500155.3	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500155	ACSL4|AFF2|AGTR2|AP1S2|ARHGEF6|ARHGEF9|ARX|ATP6AP2|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL5|CLIC2|CUL4B|DCX|DLG3|EIF2S3|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GRIA3|HCFC1|HPRT1|HSD17B10|HUWE1|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|MAGT1|MAOA|MECP2|MED12|MID1|NAA10|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|PAK3|PCDH19|PDHA1|PHF6|PHF8|PLP1|PQBP1|PRPS1|PTCHD1|RAB39B|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC6A8|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SLC9A6|SMC1A|SMS|SRPX2|SYN1|SYP|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000500162.5	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500162	ACSL4|AFF2|AGTR2|ALG6|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARHGEF6|ARHGEF9|ARID1B|ARX|ATP6AP2|ATRX|BCOR|BRWD3|C12orf57|CA8|CACNG2|CASK|CC2D1A|CCDC22|CDH15|CDKL5|CLIC2|CNTNAP2|CRBN|CTCF|CTNNB1|CUL4B|D2HGDH|DCX|DDHD2|DLG3|DYNC1H1|DYRK1A|EHMT1|EIF2S3|EPB41L1|ERLIN2|FLNA|FMR1|FOXG1|FOXP1|FRMPD4|FTSJ1|GATAD2B|GDI1|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|HCFC1|HPRT1|HSD17B10|HUWE1|IDH2|IGBP1|IL1RAPL1|IQSEC2|KCNJ10|KDM5C|KIAA2022|KIF1A|KIRREL3|KLF8|L1CAM|L2HGDH|LINS|LRP2|MAGT1|MAN1B1|MAOA|MBD5|MECP2|MED12|MED23|MEF2C|MID1|NAA10|NHS|NLGN3|NLGN4X|NRXN1|NRXN2|NSDHL|NSUN2|OCRL|OFD1|OPHN1|PACS1|PAK3|PCDH19|PCNT|PDHA1|PHF6|PHF8|PLP1|PQBP1|PRPS1|PRSS12|PTCHD1|RAB39B|RAI1|RPS6KA3|SCN2A|SHANK2|SHANK3|SHROOM4|SLC16A2|SLC25A1|SLC6A8|SLC9A6|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SMARCA4|SMC1A|SMS|SOBP|SRPX2|ST3GAL3|STXBP1|SYN1|SYNGAP1|SYP|TCF4|TECR|TRAPPC9|TSPAN7|TUBA1A|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZC3H14|ZDHHC15|ZDHHC9|ZEB2|ZNF407|ZNF41|ZNF526|ZNF674|ZNF711|ZNF81					
GTR000500166.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500166	OPHN1	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500289.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500289	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|PAFAH1B1|POMGNT1|POMT1|POMT2|RELN|TUBA1A|VLDLR	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501098.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501098	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2	Deletion/duplication analysis	Comparative Genomic Hybridization	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000501100.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501100	DCAF17	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000501110.4	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501110	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000506307.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506307	CHKB|COL6A1|COL6A2|COL6A3|DAG1|DPM2|DPM3|FKRP|FKTN|ISPD|ITGA7|LAMA2|LARGE|LMNA|POMGNT1|POMGNT2|POMT1|POMT2|RYR1|SEPN1|TMEM5	Deletion/duplication analysis	Microarray	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000506308.1	Genetic Services Laboratory	CHKB|COL6A1|COL6A2|COL6A3|DAG1|DPM2|DPM3	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	Buccal swab|Cell culture|Cord	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	University of Chicago Illinois 14D0917593 506308	|FKRP|FKTN|ISPD|ITGA7|LAMA2|LARGE|LMNA|POMGNT1|POMGNT2|POMT1|POMT2|RYR1|SEPN1|TMEM5		sequencing (MPS)	blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva		
GTR000506315.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506315	DCAF17	Deletion/duplication analysis	Microarray	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000506321.3	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506321	ANO5|CAPN3|CAV3|DAG1|DES|DNAJB6|DYSF|FKRP|FKTN|FLNC|GAA|LMNA|MYOT|PLEC|POMGNT1|POMT1|POMT2|SGCA|SGCB|SGCD|SGCG|TCAP|TRIM32|TTN	Deletion/duplication analysis	Microarray	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000506322.3	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506322	ANO5|CAPN3|CAV3|DAG1|DES|DNAJB6|DYSF|FKRP|FKTN|FLNC|GAA|LMNA|MYOT|PLEC|POMGNT1|POMT1|POMT2|SGCA|SGCB|SGCD|TCAP|TRIM32|TTN	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000506323.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506323	ATP13A2|C19orf12|CP|DCAF17|FA2H|FTL|PANK2|PLA2G6|WDR45	Deletion/duplication analysis	Microarray	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000506324.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 506324	ATP13A2|C19orf12|CP|DCAF17|FA2H|FTL|PANK2|PLA2G6|WDR45	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cell culture|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000507771.3	Genetic Services Laboratory University of Chicago Illinois 14D0917593 507771	ARFGEF2|ASPM|ATR|ATRX|CASC5|CASK|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP63|FOXG1|MCPH1|MECP2|MED17|NDE1|ORC1|ORC4|ORC6|PCNT|PNKP|RAB3GAP1|RBBP8|SLC25A19|SLC2A1|SLC9A6|STAMBP|STIL|TCF4|TSEN2|TSEN34|TSEN54|UBE3A|WDR62|ZEB2|ZNF335	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000508585.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 508585	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|GPR56|KIAA1279|LAMC3|LARGE|OCLN|PAFAH1B1|POMGNT1|POMT1|POMT2|RAB18|RAB3GAP1|RAB3GAP2|RELN|RTTN|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|WDR62	Deletion/duplication analysis	Microarray	Buccal swab|Cord blood|Fetal blood|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000508586.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 508586	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|GPR56|KIAA1279|LAMC3|LARGE|OCLN|PAFAH1B1|POMGNT1|POMT1|POMT2|RAB18|RAB3GAP1|RAB3GAP2|RELN|RTTN|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|WDR62	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cell culture|Cord blood|Fetal blood|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509252.1	Genetic Services Laboratory	CHD7|DHODH|EFTUD2|MYCN|POLR1C|POLR1D|SF	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	Amniocytes|Amniotic fluid|Buccal	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	University of Chicago Illinois 14D0917593 509252	3B4|TCOF1		sequencing (MPS)	swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva		
GTR000281976.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473 281976	MED12	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin	Decline to answer	Yes
GTR000328483.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473 328483	CASK	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin	Decline to answer	No
GTR000502023.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473	ACSL4	Mutation scanning of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole)	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	502023				blood|Saliva|Skin		
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 327662	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|HPRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No
GTR000515046.3	Illumina Clinical Services Laboratory Illumina California 05D1092911 515046	Not reported	Not reported	Not reported	Not reported	Not reported	NOT REPORTED
GTR000514956.1	Invitae — California	MED12|UPF3B	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-	Isolated DNA|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	05D2040778 514956			Generation (NGS)/Massively parallel sequencing (MPS)			
GTR000506862.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 506862	GDI1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000509437.3	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 509437	MEF2C	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500546.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500546	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500583.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	500583						
GTR000501524.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501524	ARX|CDKL5|MEF2C|SLC25A22|SRPX2|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501565.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501565	MEF2C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501566.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501566	MEF2C	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501567.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia	MEF2C	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Pennsylvania 39D0198678 501567						
GTR000501568.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501568	MEF2C	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501569.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501569	CASK	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501570.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501570	CASK	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501710.1	Molecular Genetics Laboratory Children's	SRPX2	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Hospital of Philadelphia Pennsylvania 39D0198678 501710				DNA|Paraffin block|Peripheral (whole) blood		
GTR000501714.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501714	SRPX2	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000502794.3	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 502794	FGFR2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000503333.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503333	FGFR2	Deletion/duplication analysis	Microarray	Not reported	Decline to answer	No
GTR000503358.3	Molecular Genetics	PAX6	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Children's Hospital Colorado Colorado 06D0644348 503358				swab|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Saliva		
GTR000503360.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503360	PAX6	Deletion/duplication analysis	Microarray	Not reported	Decline to answer	No
GTR000505116.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 505116	EFTUD2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000505118.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 505118	EFTUD2	Deletion/duplication analysis	Microarray	Isolated DNA|Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509671.1	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 509671	HSD17B10	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Decline to answer	Yes
GTR000507942.2	Pittsburgh Cytogenetics Laboratory University of Pittsburgh Medical Center Pennsylvania 39D0673863 507942	ABCB7|ABCD1|ACSL4|AFF2|AGTR2|AIFM1|ALAS2|ALG13|AMELX|AMER1|AP1S2|AR|ARHGEF6|ARHGEF9|ARSE|ARX|ATP2B3|ATP6AP2|ATP7A|ATRX|AVPR2|BCOR|BRWD3|BTK|CACNA1F|CASK|CDKL5|CFP|CHM|CHRDL1|CLCN5|CLIC2|COL4A5|COX7B|CSF2RA|CYBB|DCX|DKC1|DLG3|DMD|EBP|EDA|EFNB1|EMD|F8|F9|FAM58A|FANCB|FHL1|FLNA|FTSJ1|G6PD|GATA1|GDI1|GJB1|GK|GLA|GPR143|GRIA3|HCCS|HCFC1|HDAC8|HPRT1|HSD17B10|IDS|IGBP1|IGSF1|IKBKG|IL1RAPL1|IL2RG|KAL1|KDM5C|KDM6A|L1CAM|LAMP2|MAGT1|MAMLD1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NHS|NLGN3|NLGN4X|NR0B1|NSDHL|OCRL|OFD1|OPHN1|OPN1MW|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PHKA1|PHK	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniotic fluid|Cell culture|Chorionic villi|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		A2|PIGA|PLP1|POLA1|PORCN|POU3F4|PQBP1|PRPS1|RAB39B|RAB40AL|RBM10|RP2|RPGR|RPL10|RPS6KA3|RS1|SAT1|SERPINA7|SH2D1A|SHOX|SHROOM4|SLC16A2|SLC35A2|SLC6A14|SLC6A8|SLC9A6|SMPX|SMS|SOX3|SRPX2|SRY|STS|SYN1|SYP|TAF1|TAZ|TBX22|TIMM8A|TMLHE|TRAPPC2|TSPAN7|UBA1|UBE2A|UBQLN2|UPF3B|VMA21|WAS|WDR45|XIAP|XK|ZC4H2|ZDHHC15|ZDHHC9|ZIC3|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000505136.7	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505136	B3GALNT2|B3GNT1|DAG1|DPM1|DPM3|FKRP|FKTN|GMPPB|GOSR2|ISPD|LARGE|POMGNT1|POMGNT2|POMK|POMT1|POMT2|ST3GAL4|TMEM5	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000505633.4	PreventionGenetics PreventionGenetics Wisconsin	B3GALNT2|B3GNT1|CHKB|DAG1|DPM1|DPM3|FKRP|FKTN|GMPPB|GOSR2|ISPD|ITGA7|LAMA2|LARGE|LMNA|POMGNT1|POMGNT	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	52D2065132 505633	2|POMK|POMT1|POMT2|ST3GAL4|TMEM5			DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep		
GTR000505638.3	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505638	B3GALNT2|B3GNT1|DAG1|FKRP|FKTN|ISPD|LARGE|POMGNT1|POMGNT2|POMT1|POMT2|TMEM5	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000505640.4	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505640	ANO5|CAPN3|DYSF|FKRP|GMPPB|SGCA|SGCB|SGCD|SGCG|TCAP|TRAPPC11|TRIM32|TTN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep		
GTR000505780.2	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505780	FKTN	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000505832.2	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505832	FKRP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|S	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					putum|White blood cell prep		
GTR000506020.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506020	LARGE	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000506181.2	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506181	POMGNT1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000506182.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506182	POMT1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000506184.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506184	POMT2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000506404.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506404	CUL4B	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000506407.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 506407	DYNC1H1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000507440.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507440	MED12	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000507448.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507448	HEPACAM	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000507451.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507451	OPHN1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000507700.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507700	EFTUD2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000507718.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507718	TRAPPC9	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000507719.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507719	TUSC3	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000508519.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 508519	HDAC8	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000508763.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 508763	GMPPB	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509896.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 509896	ARHGEF9|ARX|CDKL5|CHD2|EFHC1|GABRA1|GABRB3|GABRG2|GRIN2A|MBD5|MECP2|MEF2C|PCDH19|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC2A1|SLC9A6|SPTAN1|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region|Targeted variant analysis	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000510109.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 510109	FGFR2	Deletion/duplication analysis|Sequence analysis of the entire coding region|Targeted variant analysis	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000501919.2	Transgenomic Transgenomic Connecticut 07D0995237	AARS2|AASS|ABAT|ABCB7|ABCD1|ABHD5|ACAD8|ACAD9|ACADL|ACADM|ACADSB|ACADVL|ACAT1|ACSF3|ACSL4|ADCK3|AFG3L	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	501919	2|AGXT|AIFM1|AIFM2|AK2|AKR1D1|ALAS2|ALDH18A1|ALDH4A1|ALDH5A1|ALDH6A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMACR|AMT|APEX2|APTX|ARG1|ARMS2|ASL|ASS1|ATL1|ATM|ATP5A1|ATP5B|ATP5C1|ATP5D|ATP5E|ATP5F1|ATP5G1|ATP5G2|ATP5G3|ATP5I|ATP5J|ATP5O|ATP7B|ATPAF1|ATPAF2|ATPIF1|ATXN10|ATXN7|AUH|B4GALT1|BCKDHA|BCKDHB|BCS1L|BRAF|BTD|C10orf2|C12orf65|C14orf2|CAPN3|CARS2|CAV3|CDKL5|CHKB|CISD2|CLN3|CLN5|CLN6|CLN8|CMC1|COG1|COG7|COG8|COQ2|COQ3|COQ4|COQ6|COQ7|COQ9|COX10|COX11|COX15|COX17|COX18|COX19|COX4I1|COX4I2|COX5A|COX5B|COX6A1|COX6A2|COX6B1|COX6C|COX7A1|COX7A2|COX7A2L|COX7B|COX7B2|COX7C|COX8A|CPOX|CPS1|CPT1A|CPT2|CRLS1|CRYAB|CTNS|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYC1|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP27A1|CYP27B1|CYP7B1|D2HGDH|DARS2|DBT|DCX|DECR1|DGUOK|DLAT|DLD|DLST|DMGDH|DMPK|DNAJC19|DNM1L|DNM2|DOLK|DPAGT1|DPM1|DPM3|EARS2|ECSIT|EIF2AK3|ELOV					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		L4|ETFA|ETFB|ETFDH|ETHE1|FA2H|FARS2|FASTKD2|FECH|FGF14|FH|FOXG1|FOXRED1|FXN|GAA|GAD1|GAMT|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLA|GLDC|GLRX5|GLUD1|GNPAT|GPD2|GPHN|HADH|HADHA|HADHB|HARS2|HAX1|HCCS|HFE|HK1|HLCS|HMGCL|HMGCS2|HSD17B10|HSD3B2|HSPD1|IARS2|IDH2|ISCU|ITPR1|IVD|KARS|KCNC3|KCNJ11|KIAA0196|KIAA0226|KIF1B|KIF5A|LARS2|LETM1|LMBRD1|LRPPRC|MAOA|MAPT|MARS2|MCCC1|MCCC2|ME2|MECP2|MFN2|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MPV17|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|MYH7|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA3|NDUFA4|NDUFA5|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAB1|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB1|NDUFB10|NDUFB11|NDUFB2|NDUFB3|NDUFB4|NDUFB5|NDUFB6|NDUFB7|NDUFB8|NDUFB9|NDUFC1|NDUFC2|NDUFS1|NDUFS2|N					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		DUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NEFL|NIPA1|NKX2-1|NPC1|NPC2|NUBPL|OAT|OGDH|OPA1|OPA3|OTC|OXA1L|OXCT1|PAFAH1B1|PANK2|PARL|PARS2|PC|PCCA|PCCB|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX13|PHB|PHYH|PLP1|PMM2|PNKD|PNPLA2|PNPLA3|POLG|POLG2|PPM1B|PPOX|PPT1|PREPL|PRKCG|PRODH|PUS1|PWAR1|RARS2|REEP1|RFT1|RMRP|RNASEH2A|RNASEH2B|RNASEH2C|RRM2B|RYR1|SAMHD1|SARS2|SCN1A|SCO1|SCO2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SGCD|SLC12A3|SLC16A2|SLC19A2|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A16|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC29A3|SLC2A1|SLC2A10|SLC33A1|SLC35A1|SLC35C1|SLC3A1|SLC52A1|SLC6A8|SLC7A9|SOD1|SPAST|SPG11|SPG20|SPG7|SPTBN2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TACO1|TARS2|TAZ|TIMM8A|TK2|TMEM126A|TMEM70|TOP1MT|TPM2|TPP1|TRMU|TSFM|TTBK2|TUFM|TYMP|UBE3A|UCP1|UCP2|U					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CP3|UNG|UQCR10|UQCR11|UQCRB|UQCRC1|UQCRC2|UQCRFS1|UQCRH|UQCRQ|UROS|USMG5|VARS2|WARS2|WFS1|XDH|XPNPEP3|YARS2|ZFYVE26					
GTR000509336.2	Transgenomic Transgenomic Connecticut 07D0995237 509336	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FL	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000245064.4	University of Iowa Diagnostic	Not reported	Sequence analysis of the entire coding	Bi-directional Sanger Sequence Analysis|PCR	Cell culture|Peripheral	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 245064		region|Targeted variant analysis	with allele specific hybridization	(whole) blood		
GTR000322431.2	University of Iowa Diagnostic Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 322431	POMT1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Cell culture|Peripheral (whole) blood	Decline to answer	Yes
GTR000325670.1	University of Iowa Diagnostic Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 325670	FKTN	Sequence analysis of the entire coding region|Targeted variant analysis	Bi-directional Sanger Sequence Analysis|PCR with allele specific hybridization	Cell culture|Peripheral (whole) blood	Decline to answer	Yes
GTR000330743.2	University of Iowa Diagnostic Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 330743	LARGE	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Cell culture|Peripheral (whole) blood	Decline to answer	Yes

Table D-6. Genetic tests for Prader-Willi syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000335345.1	BayCare Cytogenetics Laboratory BayCare Laboratories, LLC Florida 10D2029560 335345	SNRPN	FISH-metaphase	Digital/Virtual karyotyping	Peripheral (whole) blood	Decline to answer	Yes
GTR000512115.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512115	15q11-q13	Targeted variant analysis	Trinucleotide repeat by PCR or Southern Blot	Not reported	Not reported	No
GTR000502862.1	Center for Genetics at Saint Francis Saint Francis Hospital Oklahoma 37D0474681 502862	SNRPN	Methylation analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000502968.1	Center for Genetics at Saint Francis Saint Francis Hospital Oklahoma 37D0474681	SNRPN	Methylation analysis	Methylation-specific PCR	Cell culture|Isolated DNA|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	502968						
GTR000006792.2	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 6792	SNRPN, 15q11.2, Chromsome 15	FISH-metaphase|Methylation analysis|Uniparental disomy study (UPD)	Other|Methylation-specific PCR|Other	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000206885.1	Center for Human Genetics, Inc Massachusetts 22D0650242 206885	15q11-q13	FISH-metaphase|Deletion/duplication analysis|Methylation analysis|Uniparental disomy study (UPD)	Fluorescence in situ Hybridization (FISH)|Multiplex Ligation-dependent Probe Amplification (MLPA)|Multiplex Ligation-dependent Probe Amplification (MLPA)|PCR on ABI	Not reported	Decline to answer	No
GTR000196494.4	ChildLab Molecular Genetics Laboratory Nationwide Children's Hospital Ohio 36D0665271 196494	SNRPN, 15q11.2-q13	FISH-metaphase|Methylation analysis|Uniparental disomy study (UPD)	FISH|Methylation-specific PCR|characterization of microsatellite polymorphisms on an ABI instrument	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000295247.2	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 295247	15q11.2-q13	FISH-metaphase	Metaphase FISH analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000509337.1	Clinical Molecular Genetics Laboratory All Children's Hospital Florida 10D0700790 509337	15q11-q13	Methylation analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Buccal swab|Isolated DNA|Peripheral (whole) blood	No	Yes
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000327603.1	Cytogenetics and Molecular Diagnostics Lab CGC Genetics USA New Jersey 31D1085261 327603	Prader-Willi critical region	FISH-interphase|FISH-metaphase|Deletion/duplication analysis|Methylation analysis	FISH|FISH|Multiplex Ligation-dependent Probe Amplification (MLPA)|Methylation-specific PCR	Cell-free DNA|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000325591.1	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center	15q11.2	FISH-metaphase|Methylation analysis	FISH|Methylation-specific PCR	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Ohio 36D0965797 325591						
GTR000509082.1	Cytogenetics and Molecular Pathology Laboratory DLP Marquette General Hospital, LLC Michigan 23D1062707 509082	PWARSN	FISH-interphase|FISH-metaphase|Karyotyping	FISH|FISH|G-banding	Buccal swab|Cord blood|Fetal blood|Peripheral (whole) blood	No	Yes
GTR000013522.1	Cytogenetics Laboratory Indiana University School of Medicine Indiana 15D0647198 13522	15q11.2-q13	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization (FISH)|Fluorescence in situ hybridization (FISH)	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin|Urine	Decline to answer	Yes
GTR000032599.1	Cytogenetics Laboratory SUNY Upstate Medical University New York 33D0654590 32599	15q11.2	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Product of conception (POC)|Skin	Yes	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500042.2	Cytogenetics Laboratory ARUP Laboratories, Inc. Utah 46D0523979 500042	15q11.2-q13|17p11.2|17p13.3|22q11.2|22q13.3|4p16.3|5p15.2|7q11.23|Xp22.3|Yp11.3	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000503269.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503269	SNRPN	Methylation analysis	Methylation-specific PCR	Not reported	No	No
GTR000503277.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 503277	DMPK|MEG3|SMN1|SNRPN	Methylation analysis|Targeted variant analysis	Methylation-specific PCR|Allele-specific PCR|Trinucleotide repeat by PCR or Southern Blot	Not reported	No	No
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		FAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2					
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509392.9	Fulgent Clinical Diagnostics Lab	ABCC8|ALMS1|ARL6|BBS1|BBS10|BBS12|BBS2|BBS	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Buccal swab|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509392	4|BBS5|BBS7|BBS9|BDNF|CEL|CEP290|EIF2AK3|FOXP3|GCK|GLIS3|GNAS|HNF1A|HNF1B|HNF4A|INS|KCNJ11|LEP|LEPR|MAGEL2|MC4R|MKKS|MKS1|NEUROD1|NEUROG3|NTRK2|PCSK1|PDX1|POMC|PTF1A|RFX6|SDCCAG8|SIM1|TRIM32|TTC8|WDPCP|WFS1	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	(whole) blood|Saliva		
GTR000006812.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 6812	15q11.2-q13	Methylation analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500159.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500159	15q11-q13	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509731.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 509731	MAGEL2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000274682.3	Genetics Laboratory University of Oklahoma Health Sciences Center Oklahoma 37D0967945 274682	SNRPN	Methylation analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000500313.1	Genetics Laboratory Shodair Children's Hospital Montana 27D0652530 500313	SNRPN, 15q11.2-q13	FISH-metaphase|Methylation analysis|Uniparental disomy study (UPD)	FISH|Methylation-specific PCR|Trinucleotide repeat by PCR or Southern Blot	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000004542.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center	15q11-q13	Methylation analysis|Uniparental disomy study (UPD)	Pyrosequencing|Other	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	South Carolina 42D0689473 4542				DNA|Peripheral (whole) blood|Saliva|Skin		
GTR000053126.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 53126	15q11.2-q13	FISH-interphase|FISH-metaphase|Methylation analysis|Uniparental disomy study (UPD)	Fluorescence in situ hybridization|Fluorescence in situ hybridization|Methylation-specific PCR|Other	Fetal blood|Peripheral (whole) blood	No	Yes
GTR000006562.2	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 6562	SNRPN	Deletion/duplication analysis|Methylation analysis	MS-MLPA analysis of undigested and digested genomic DNA|MS-MLPA analysis of undigested and digested genomic DNA	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000309920.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 309920	SNRPN	Methylation analysis	Methylation-specific PCR	Not reported	Decline to answer	Yes
GTR000500630.2	Michigan State University Clinical	SNRPN	Methylation analysis	PCR with allele specific hybridization	Bone marrow|Peripheral	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratory Michigan State University Michigan 23D0650879 500630				(whole) blood		
GTR000500648.2	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500648	15q11.2	FISH-metaphase	Other	Amniocytes|Bone marrow|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	No	Yes
GTR000005898.3	Molecular and Biochemical Genetics Laboratory Dayton Children's Hospital Ohio 36D0859317 5898	SNRPN	Methylation analysis	Methylation-specific PCR	Isolated DNA	Decline to answer	Yes
GTR000084935.1	Molecular Diagnostic Laboratory Barnes Jewish Hospital Missouri 26D0438670 84935	SNRPN	Methylation analysis	PCR-RFLP with Southern hybridization	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000253215.1	Molecular Diagnostics Laboratory Duke University Health System North Carolina 34D1005312 253215	SNRPN	Methylation analysis	Methylation-specific PCR	Buccal swab|Peripheral (whole) blood	Decline to answer	Yes
GTR000500322.1	Molecular Diagnostics Laboratory University of California, San Francisco California 05D1024215 500322	SNRPN	Methylation analysis	Other	Cell culture|Peripheral (whole) blood	No	Yes
GTR000336252.1	Molecular Genetics Diagnostic Laboratory Detroit Medical Center University Laboratories Michigan 23D0717194 336252	SNRPN	Methylation analysis	Methylation-specific PCR	Peripheral (whole) blood	Decline to answer	Yes
GTR000166462.2	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania	SNRPN	Methylation analysis	Methylation-specific PCR	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	39D0198678 166462						
GTR000193079.1	Molecular Genetics Laboratory Children's Mercy Hospital and Clinics Missouri 26D2046586 193079	15q11.2-q13	Deletion/duplication analysis|Methylation analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)|Methylation-specific PCR	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000204881.2	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 204881	SNRPN	Methylation analysis	Methylation-specific PCR	Not reported	Decline to answer	Yes
GTR000500706.2	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500706	SNRPN	Methylation analysis	Methylation-specific PCR|Other	Peripheral (whole) blood	Decline to answer	Yes
GTR000503131.2	Natera California 05D1082992 503131	15q11-q13|1p36|22q11.2|5p|Chromosome 13|Chromosome 18|Chromosome 21|Human genome|Sex chromosome	Targeted variant analysis	SNP Detection	Cell-free DNA|Maternal blood sample	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		X					
GTR000500554.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 500554	SNRPN	Methylation analysis	Methylation-specific PCR	Not reported	No	Yes
GTR000500555.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 500555	SNRPN	Methylation analysis	Methylation-specific PCR	Not reported	No	Yes
GTR000504363.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504363	15q11.2	FISH-metaphase	Other	Peripheral (whole) blood	No	Yes
GTR000506046.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506046	15q11.2-q13	FISH-metaphase	Other	Amniocytes|Chorionic villi|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501985.3	Sequenom Center for Molecular Medicine - San Diego Sequenom CMM California 05D2015356 501985	1p36|22q11.2|5p15.2|Chromosome 13|Chromosome 15|Chromosome 16|Chromosome 18|Chromosome 21|Chromosome 22|Sex chromosome X|Sex chromosome Y	Targeted variant analysis	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes
GTR000025316.1	United States Air Force, DNA Diagnostic Laboratory United States Air Force Mississippi 25316	Prader-Willi critical region	Methylation analysis	Methylation-specific PCR	Peripheral (whole) blood	Decline to answer	No
GTR000219915.1	University of Iowa Diagnostic Laboratories University of Iowa Hospitals and Clinics Iowa 16D0664625 219915	SNRPN, 15q11.2-q13	Deletion/duplication analysis|Methylation analysis	Southern blot hybridization|Southern blot hybridization	Cell culture|Peripheral (whole) blood	Decline to answer	Yes
GTR000305781.1	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305781	15q11-q13	FISH-interphase|FISH-metaphase|Deletion/duplication analysis|Methylation analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray|Methylation-specific PCR	Amniocytes|Amniotic fluid|Cord blood|Peripheral (whole) blood	Decline to answer	Yes

Table D-7. Genetic tests for Rett syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000305902.5	(AC) Atlanta Center: Medical Neurogenetics, LLC. NR Georgia 11D0703390 305902	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Buccal swab|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000319372.1	Ambry Genetics Ambry Genetics California 05D0981414 319372	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000331025.2	Ambry Genetics Ambry Genetics California 05D0981414 331025	CDKL5|MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Peripheral (whole) blood|Saliva	No	Yes
GTR000500399.2	Ambry Genetics Ambry Genetics California 05D0981414 500399	FOXG1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000500401.2	Ambry Genetics Ambry Genetics California 05D0981414 500401	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Cell culture|Chorionic villi|Isolated DNA|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500402.2	Ambry Genetics Ambry Genetics California 05D0981414 500402	CDKL5|FOXG1|MECP2|MEF2C	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Cell culture|Chorionic villi|Cord blood|Isolated DNA|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000512790.2	Athena Diagnostics Inc NR Massachusetts 22D0069726 512790	ARHGEF9|ARX|ATP6AP2|ATRX|CASK|CDKL5|CUL4B|DCX|FGD1|GPC3|GRIA3|HSD17B10|KDM5C|MECP2|OFD1|OPHN1|PAK3|PCDH19|PHF6|PLP1|PQBP1|RAB39B|SLC9A6|SMC1A|SMS|SRPX2|SYP	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000512791.2	Athena Diagnostics Inc NR Massachusetts 22D0069726 512791	ARHGEF9|ARX|CDKL5|CNTNAP2|FOXG1|GABRG2|GRIN2A|KCNT1|MECP2|NRXN1|PCDH19|PNKP|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYNGAP1|TCF4|TREX1|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000512792.2	Athena Diagnostics Inc NR Massachusetts 22D0069726 512792	ARX|CDKL5|FOXG1|GABRB3|GRIN2A|MEF2C|SCN2A|SLC25A22|SPTAN1|STXBP1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000512796.2	Athena Diagnostics Inc NR Massachusetts	ALDH7A1|ARFGEF2|ARHGEF9|ARX|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATRX|CACNA1A|CASK|CASR|CCDC88C|CDKL5|CHRNA	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	22D0069726 512796	2|CHRNA4|CHRNB2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COL18A1|COL4A1|CPT2|CSTB|CTSD|CUL4B|DCX|DEPDC5|DNAJC5|EFHC1|EMX2|EPM2A|FGD1|FGFR3|FKRP|FKTN|FLNA|FOXG1|GABRA1|GABRB3|GABRD|GABRG2|GPC3|GPR56|GRIA3|GRIN2A|HSD17B10|KCNA1|KCNJ1|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|LAMA2|LARGE|LBR|LGI1|MBD5|ME2|MECP2|MEF2C|MFSD8|NHLRC1|NIPBL|NOTCH3|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PCDH19|PEX7|PHF6|PIGV|PLA2G6|PLP1|PNKP|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRRT2|RAB39B|RAB3GAP1|RAI1|RELN|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SERPINI1|SETBP1|SLC25A22|SLC2A1|SLC4A10|SLC9A6|SMC1A|SMC3|SMS|SNAP29|SPTAN1|SRPX2|STXBP1|SYNGAP1|SYP|TBC1D24|TBX1|TCF4|TPP1|TREX1|TSC1|TSC2|TUBA1A|TUBA8|TUBB2B|UBE3A|VPS13A|VPS13B|WDR62|ZEB2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000515153.1	Athena Diagnostics Inc NR Massachusetts 22D0069726 515153	FOXG1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000515156.1	Athena Diagnostics Inc NR Massachusetts 22D0069726 515156	FOXG1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000515165.1	Athena Diagnostics Inc NR Massachusetts 22D0069726 515165	FOXG1	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	NR	No	Yes
GTR000515172.1	Athena Diagnostics Inc NR Massachusetts 22D0069726 515172	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000515179.1	Athena Diagnostics Inc NR Massachusetts 22D0069726 515179	MECP2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000515187.1	Athena	MECP2	Deletion/duplication	Multiplex Ligation-dependent	NR	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Inc NR Massachusetts 22D0069726 515187		analysis	Probe Amplification (MLPA)			
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|ALDOA|ALDOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CACNA2D4|CASP8|CAT|CBS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM1L|DPM1|DPM3|DSP|DTNBP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCTK|GM2A|GNAT1|GNAT2|GNE|GNPTAB|GNS|GOT1|GPD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|				

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|MC1R|MCCC1|MCCC2|MCEE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NDUFV2|NEFH|NEUROD1|NFU1|NHS|NME1|NPHP1|NPHP3|NPHP4|NRL|NT5C3A|NUBPL|NYX|OAT|OCA2|OCRL|OGDH|OGG1|OPA1|OPA3|OPN1LW|OPN1MW|OPTN|OSTM1|OTC|OTX2|OXCT1|PAH|PANK2|PARK2|PARK7|PAX2|PAX6|PC|PCCA|PCCB|PCDH15|PCK2|PDE6A|PDE6B|PDE6C|PDE6G|PDHA1|PDHB|PDP1|PDSS1|PDSS2|PDZD7|PFKM|PGAM2|PGM1|PHB|PHKA1|PHKA2|PHKB|PHKG2|PHOX2A|PHYH|PINK1|PITPNM3|PITX2|PITX3|PLA2G2A|PLOD2|PLOD3|PMM2|PNKD|PNPLA2|POLG|POLG2|PPARG|PPARGC1B|PPIB|PPOX|PPP2R1B|PRCD|PRKCG|PRODH|PROM1|PRPF31|PRPF6|PRPF8|PRPH2|PSAP|PSEN1|PTRF|PTS|PUS1|PYCR1|PYGL|PYGM|QDPR|RAF1|RARS2|RAX|RB1|RD3|RDH12|RDH5|REEP1|RET|RFT1|RGR|RGS9|RHO|RILP|RIMS1|RLBP1|RNASEL|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPL35A|RPS14|RRM2B|RS1|SAG|SARDH|SARS2|SCO2|SCP2|SDHAF1|SDHAF2|SDHB|SDHC|SECISBP2|SEMA4A|SEPT9|SERPINF1|SGSH|SHH|SIX6|SLC16A1|SLC22A4|SLC22A5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|TRPM1|TSFM|TSPAN12|TTC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513				
GTR000511961.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511961	FOXG1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	NR	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511962.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511962	FOXG1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000511963.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511963	FOXG1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512051.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512051	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512052.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512052	MECP2	Deletion/duplication analysis	Trinucleotide repeat by PCR or Southern Blot	NR	NR	No
GTR000512053.1	Baylor Medical	MECP2	Deletion/duplication	Trinucleotide repeat by PCR	NR	NR	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512053		analysis	or Southern Blot			
GTR000512054.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512054	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512055.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512055	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000512056.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512056	MECP2	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	NR	NR	No
GTR000026276.1	Center for Human Genetics,	MECP2	Deletion/duplication analysis|Sequence analysis	Multiplex Ligation-dependent Probe Amplification	NR	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Inc NR Massachusetts 22D0650242 26276		of the entire coding region	(MLPA)|Bi-directional Sanger Sequence Analysis			
GTR000298451.1	Center for Human Genetics, Inc NR Massachusetts 22D0650242 298451	Xq28	FISH-interphase|FISH-metaphase|Deletion/duplication analysis	Fluorescence in situ Hybridization (FISH)|Fluorescence in situ Hybridization (FISH)|Multiplex Ligation-dependent Probe Amplification (MLPA)	NR	Decline to answer	No
GTR000322959.1	Center for Human Genetics, Inc NR Massachusetts 22D0650242 322959	FOXG1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	NR	Decline to answer	No
GTR000509362.2	Clinical Molecular Genetics Laboratory All Children's Hospital Florida 10D0700790 509362	MECP2	Mutation scanning of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Buccal swab|Chorionic villi|Fibroblasts|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	No
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AM	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Produ	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	22D2035168 500883	T|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|BRD2|BTD|BUB1B|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|			ct of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDH2|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000500936.1	Courtagen Diagnostics	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|AB	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	Cord blood|Fresh tissue|Frozen	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500936	CC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|G		sequencing (MPS)	tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 503671	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXR	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000510912.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 510912	ADSL|ALDH7A1|ARHGEF9|ARX|ATP6AP2|CACNB4|CDKL5|CHD2|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CSTB|CTSD|EFHC1|EPM2A|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|GATM|GOSR2|GRIN2A|KANSL1|KCNJ10|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MECP2|MEF2C|MFSD8|NHLRC1|NRXN1|PCDH19|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRRT2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC25A22|SLC2A1|SLC9A6|SPTAN1|STXBP1|SYN1|SYNGAP1|TBC1D24|TCF4|TPP1|TSC1|TSC2|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fetal blood|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000501512.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501512	FOXG1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501513.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501513	FOXG1	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	No	No
GTR000502819.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502819	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	No	No
GTR000502820.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502820	MECP2	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	No	No
GTR000512416.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512416	ABAT|ADSL|ALDH7A1|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|BCKDK|CACNA1A|CACNB4|CASK|CASR|CDKL5|CENPJ|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DNAJC5|EFHC1|EMX2|EPM2A|FLNA|FOLR1|FOXG1|GABRA1|GABRG2|GAMT|G	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ATM|GOSR2|GPR56|GPR98|GRIN2A|HCN1|HCN4|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|LGI1|LIAS|MAGI2|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MTHFR|NDE1|NDUFA1|NHLRC1|NRXN1|OPHN1|PAFAH1B1|PCDH19|PHF6|PLCB1|PNKP|PNPO|POLG|PPT1|PRICKLE1|PRICKLE2|PRRT2|RELN|SCARB2|SCN1A|SCN1B|SCN2A|SCN3A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TPP1|TSC1|TSC2|TSEN54|UBE3A|WDR62|ZEB2					
GTR000512586.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512586	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK16|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|HCFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|P	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		DHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711					
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No
GTR000512608.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512608	ACTB|ACTG1|ADSL|AHI1|ALDH7A1|ARFGEF2|ARHGEF9|ARX|ASPM|ATP1A2|ATP6AP2|ATR|ATRX|BCKDK|CACNB4|CASC5|CASK|CC2D2A|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP290|CEP41|CEP63|CHMP1A|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|D	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CX|DHCR7|DISP1|DNAJC5|EFHC1|EHMT1|EOMES|EPM2A|EXOSC3|FGF8|FKRP|FKTN|FLNA|FOLR1|FOXG1|FOXH1|GABRA1|GABRG2|GAMT|GATM|GLI2|GOSR2|GPR56|GPR98|GRIN2A|GRIN2B|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KIAA1279|KIF7|LAMC3|LARGE|LARP7|LGI1|LIAS|MAGI2|MAPK10|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MKS1|MYCN|NDE1|NHLRC1|NIN|NODAL|NPHP1|NRXN1|OPHN1|ORC4|ORC6|PAFAH1B1|PCDH19|PCNT|PLCB1|PNKP|PNPO|POC1A|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRRT2|PTCH1|RAB18|RAB3GAP1|RAB3GAP2|RARS2|RBBP8|RELN|RPGRIP1L|RTTN|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TGIF1|TMEM138|TMEM216|TMEM237|TMEM67|TPP1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|UBE3A|VLDLR|VRK1|WDR62|ZEB2|ZIC2|ZNF335					
GTR000509342.9	Fulgent Clinical Diagnostics Lab	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Cell culture|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509342	BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	(whole) blood|Saliva		
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509399	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000509430.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509430	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|LARGE|PAFAH1B1|POMGNT1|POMT1|POMT2|RELN|TUBA1A|VLDLR	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509432.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509432	ACAD9|ADCK3|APTX|ATP5E|ATPAF2|BCS1L|COQ2|COQ9|COX10|COX15|COX6B1|DARS2|DGUOK|DLAT|DLD|DNM1L|ETFA|ETFB|ETFDH|ETHE1|FBP1|FH|FOXRED1|G6PC|GFM1|GYS2|ISCU|LRPPRC|MRPS16|MRPS22|NDUFA11|NDUFAF1|NDUFAF3|NDUFAF4|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|PC|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|POLG|PUS1|RRM2B|SCO2|SLC25A3|SLC25A4|SUCLA2|SUCLG1|TAZ|TK2|TMEM70|TRMU|TSFM|TUFM|TYMP|UQCRB|YARS2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509436.8	Fulgent Clinical Diagnostics Lab	ABCC8|ACY1|ADAMTSL2|ADSL|AGA|ALDH4A1|ALD	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Buccal swab|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509436	H5A1|ALDH7A1|AMT|ANTXR2|ARG1|ARSA|ARSB|ASAH1|ASPA|ATP13A2|BTD|CLN3|CLN5|CLN6|CLN8|COL11A2|COL2A1|CTNS|CTSA|CTSC|CTSD|CTSK|DHCR7|DNAJC5|DPYD|DYM|ETFA|ETFB|ETFDH|FH|FOLR1|FUCA1|GAA|GALC|GALNS|GAMT|GBA|GCDH|GCSH|GLA|GLB1|GLDC|GM2A|GNE|GNPTAB|GNPTG|GNS|GPC3|GUSB|HEXA|HEXB|HGSNAT|HPD|HRAS|IDS|IDUA|L2HGDH|LAMA2|LAMP2|LIPA|LMBRD1|MAN2B1|MANBA|MCOLN1|MFSD8|MOCS1|MOCS2|NAGA|NAGLU|NEU1|NPC1|NPC2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX26|PEX3|PEX5|PEX6|PGK1|PHYH|PPT1|PRODH|PSAP|QDPR|RAI1|SGSH|SLC17A5|SLC25A15|SLC46A1|SMPD1|SUMF1|SUOX|TCF4|TPP1	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	(whole) blood|Saliva		
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509442	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AV	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAM					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOB					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		P|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509443.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509443	ALDH7A1|ARX|ATP1A2|CACNA1A|CDKL5|FOLR1|FOXG1|GAMT|KCNQ2|MECP2|PCDH19|PHGDH|PNPO|POLG|PPT1|SCN1A|SLC2A1|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509444.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509444	ACAD9|ACADL|ACADM|ACADVL|AGL|C10orf2|CPT1B|CPT2|GAA|GYS1|HADHA|HADHB|OPA1|OPA3|PFKM|PGAM2|PGM1|PHKA1|POLG|POLG2|RRM2B|SUCLA2|TK2|TYMP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509446.8	Fulgent Clinical Diagnostics Lab	C10orf2|DGUOK|MPV17|POLG|POLG2|RRM2B|SLC2	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Buccal swab|Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509446	5A4|SUCLA2|SUCLG1|TK2|TYMP	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	(whole) blood|Saliva		
GTR000509453.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509453	CLN3|CLN5|CLN6|CLN8|CTSD|DNAJC5|KCTD7|MFSD8|PPT1|TPP1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509462.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509462	ARX|ATRX|CAV1|CDKL5|CNTNAP2|FOXG1|MECP2|MED17|MEF2C|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|TRAPPC9|UBE3A|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510870.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510870	DHFR|FOLR1|FPGS|MTHFD1|MTHFD1L|MTHFS|PTS|SHMT1|SLC19A1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACS	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	510915	M3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKDHA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|CLYBL|CNR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYCTK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000510916.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510916	ABCD1|ACSL4|AFF2|AGTR2|AIFM1|AP1S2|ARHGEF6|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|EBP|FAAH2|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|GSPT2|HCCS|HDAC8|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|L1CAM|LAMP2|MAGT1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTC	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		HD1|RAB39B|RAB40AL|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SRPX2|SYN1|SYP|TAF1|TIMM8A|TSPAN7|UBE2A|UPF3B|USP9X|WDR13|ZCCHC12|ZDHHC15|ZDHHC9|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000055238.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 55238	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000298455.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 298455	MECP2	Deletion/duplication analysis	Methylation-specific PCR	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000319559.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 319559	FOXG1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500160.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500160	CDKL5|FOXG1|MECP2|MEF2C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500165.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500165	FOXG1	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000501098.2	Genetic Services Laboratory	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR	Deletion/duplication analysis	Comparative Genomic Hybridization	Isolated DNA|Peripheral	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	University of Chicago Illinois 14D0917593 501098	1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2			(whole) blood		
GTR000501110.4	Genetic Services Laboratory University of Chicago Illinois 14D0917593 501110	ARX|ATRX|CDKL5|CNTNAP2|DYRK1A|EHMT1|FOLR1|FOXG1|MBD5|MECP2|MEF2C|NRXN1|OPHN1|PCDH19|PNKP|SLC2A1|SLC9A6|TCF4|UBE3A|ZEB2	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000507771.3	Genetic Services Laboratory University of Chicago Illinois 14D0917593 507771	ARFGEF2|ASPM|ATR|ATRX|CASC5|CASK|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP63|FOXG1|MCPH1|MECP2|MED17|NDE1|ORC1|ORC4|ORC6|PCNT|PNKP|RAB3GAP1|RBBP8|SLC25A19|SLC2A1|SLC9A6|STAMBP|STIL|TCF4|TSEN2|TSEN34|TSEN54|UBE3A|WDR62|ZEB2|ZNF335	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000321412.1	Genetics Laboratory University of Oklahoma Health Sciences Center Oklahoma 37D0967945 321412	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000030362.1	Greenwood Genetic Center Diagnostic	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger	Amniocytes|Amniotic fluid|Cell culture|Chorionic	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratories Greenwood Genetic Center South Carolina 42D0689473 30362			Sequence Analysis	villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin		
GTR000314263.1	Greenwood Genetic Center Diagnostic Laboratories Greenwood Genetic Center South Carolina 42D0689473 314263	FOXG1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Isolated DNA|Peripheral (whole) blood|Saliva|Skin	Decline to answer	Yes
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 327662	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|HPRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2					
GTR000004996.2	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 4996	MECP2	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000232787.2	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon 38D0881787 232787	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000331373.2	Knight Diagnostic Laboratories - Molecular Diagnostic Center Oregon Health and Science University Oregon	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Fetal blood|Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	38D0881787 331373						
GTR000205558.4	Molecular Diagnostics Lab Nemours Alfred I. duPont Hospital for Children Delaware 08D0706140 205558	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Quantitative multiplex PCR to determine copy number|Bi-directional Sanger Sequence Analysis	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000328852.3	Molecular Diagnostics Lab Nemours Alfred I. duPont Hospital for Children Delaware 08D0706140 328852	MECP2	Deletion/duplication analysis	Quantitative multiplex PCR to determine exon copy number	Isolated DNA|Peripheral (whole) blood	Decline to answer	Yes
GTR000322184.2	Molecular Genetics Diagnostic Laboratory Detroit Medical Center University Laboratories Michigan 23D0717194 322184	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000245100.1	Molecular Genetics Laboratory Children's Mercy Hospital and Clinics	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Isolated DNA|Peripheral (whole) blood	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Missouri 26D2046586 245100						
GTR000298463.1	Molecular Genetics Laboratory Children's Mercy Hospital and Clinics Missouri 26D2046586 298463	MECP2	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Isolated DNA|Peripheral (whole) blood	Decline to answer	No
GTR000500546.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500546	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500583.1	Molecular Genetics Laboratory ARUP Laboratories Utah 46D0523979 500583	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood	Decline to answer	Yes
GTR000500584.1	Molecular Genetics Laboratory ARUP Laboratories	MECP2	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Utah 46D0523979 500584						
GTR000501476.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501476	FOXG1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501477.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501477	FOXG1	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501478.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501478	FOXG1	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501479.1	Molecular Genetics Laboratory Children's	FOXG1	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Hospital of Philadelphia Pennsylvania 39D0198678 501479				DNA|Paraffin block|Peripheral (whole) blood		
GTR000501562.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501562	ARX|CDKL5|MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501700.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501700	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501701.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501701	MECP2	Sequence analysis of the entire coding region	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501702.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501702	MECP2	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000501703.1	Molecular Genetics Laboratory Children's Hospital of Philadelphia Pennsylvania 39D0198678 501703	MECP2	Targeted variant analysis	Uni-directional Sanger sequencing	Bone marrow|Buccal swab|Cell culture|Fibroblasts|Frozen tissue|Isolated DNA|Paraffin block|Peripheral (whole) blood	No	Yes
GTR000507942.2	Pittsburgh Cytogenetics Laboratory University of Pittsburgh Medical Center Pennsylvania 39D0673863 507942	ABCB7|ABCD1|ACSL4|AFF2|AGTR2|AIFM1|ALAS2|ALG13|AMELX|AMER1|AP1S2|AR|ARHGEF6|ARHGEF9|ARSE|ARX|ATP2B3|ATP6AP2|ATP7A|ATRX|AVPR2|BCOR|BRWD3|BTK|CACNA1F|CASK|CDKL5|CFP|CHM|CHRDL1|CLCN5|CLIC2|COL4A5|COX7B|CSF2RA|CYBB|DCX|DKC1|DLG3|DMD|EBP|EDA|EFNB1|EMD|F8|F9|FAM58A|FANCB|FHL1|FLNA|FTSJ1|G6PD|GATA1|GDI1|GJB1|GK|GLA|GPR143|GRIA3|HCCS|HCFC1|HDAC8|HPRT1|HSD17B10|IDS|IGBP1|IGSF1|IKBKG|IL1RAPL1|IL2RG|KAL1|KD	Deletion/duplication analysis	Comparative Genomic Hybridization	Amniotic fluid|Cell culture|Chorionic villi|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		M5C|KDM6A|L1CAM|LAMP2|MAGT1|MAMLD1|MAOA|MBTPS2|MECP2|MED12|MID1|MTM1|NAA10|NDP|NHS|NLGN3|NLGN4X|NR0B1|NSDHL|OCRL|OFD1|OPHN1|OPN1MW|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PHKA1|PHKA2|PIGA|PLP1|POLA1|PORCN|POU3F4|PQBP1|PRPS1|RAB39B|RAB40AL|RBM10|RP2|RPGR|RPL10|RPS6KA3|RS1|SAT1|SERPINA7|SH2D1A|SHOX|SHROOM4|SLC16A2|SLC35A2|SLC6A14|SLC6A8|SLC9A6|SMPX|SMS|SOX3|SRPX2|SRY|STS|SYN1|SYP|TAF1|TAZ|TBX22|TIMM8A|TMLHE|TRAPPC2|TSPAN7|UBA1|UBE2A|UBQLN2|UPF3B|VMA21|WAS|WDR45|XIAP|XK|ZC4H2|ZDHHC15|ZDHHC9|ZIC3|ZNF41|ZNF674|ZNF711|ZNF81					
GTR000508539.3	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 508539	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					(POC)|Saliva|Skin|Sputum|White blood cell prep		
GTR000509896.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 509896	ARHGEF9|ARX|CDKL5|CHD2|EFHC1|GABRA1|GABRB3|GABRG2|GRIN2A|MBD5|MECP2|MEF2C|PCDH19|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SLC2A1|SLC9A6|SPTAN1|STXBP1	Deletion/duplication analysis|Sequence analysis of the entire coding region|Targeted variant analysis	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes
GTR000511097.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 511097	FOXG1	Sequence analysis of the entire coding region|Targeted variant analysis	Bi-directional Sanger Sequence Analysis|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000508076.1	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 508076	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	No	Yes
GTR000508077.1	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 508077	MECP2	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	No	Yes
GTR000508078.1	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 508078	MECP2	Deletion/duplication analysis	Other	NR	No	Yes
GTR000336125.2	Transgenomic Transgenomic Connecticut 07D0995237 336125	MECP2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA)|Bi-directional Sanger Sequence Analysis	Peripheral (whole) blood|Saliva	Decline to answer	Yes
GTR000501919.2	Transgenomic Transgenomic Connecticut	AARS2|AASS|ABAT|ABCB7|ABCD1|ABHD5|ACAD8|ACAD9|ACADL|ACADM|ACADSB|ACADVL|ACAT1|AC	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	07D0995237 501919	SF3|ACSL4|ADCK3|AFG3L2|AGXT|AIFM1|AIFM2|AK2|AKR1D1|ALAS2|ALDH18A1|ALDH4A1|ALDH5A1|ALDH6A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMACR|AMT|APEX2|APTX|ARG1|ARMS2|ASL|ASS1|ATL1|ATM|ATP5A1|ATP5B|ATP5C1|ATP5D|ATP5E|ATP5F1|ATP5G1|ATP5G2|ATP5G3|ATP5I|ATP5J|ATP5O|ATP7B|ATPAF1|ATPAF2|ATPIF1|ATXN10|ATXN7|AUH|B4GALT1|BCKDHA|BCKDHB|BCS1L|BRAF|BTD|C10orf2|C12orf65|C14orf2|CAPN3|CARS2|CAV3|CDKL5|CHKB|CISD2|CLN3|CLN5|CLN6|CLN8|CMC1|COG1|COG7|COG8|COQ2|COQ3|COQ4|COQ6|COQ7|COQ9|COX10|COX11|COX15|COX17|COX18|COX19|COX4I1|COX4I2|COX5A|COX5B|COX6A1|COX6A2|COX6B1|COX6C|COX7A1|COX7A2|COX7A2L|COX7B|COX7B2|COX7C|COX8A|CPOX|CPS1|CPT1A|CPT2|CRLS1|CRYAB|CTNS|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYC1|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP27A1|CYP27B1|CYP7B1|D2HGDH|DARS2|DBT|DCX|DECR1|DGUOK|DLAT|DLD|DLST|DMGDH|DMPK|DNAJC19|DNM1L|DNM2|DOLK|DPAGT1|DPM1|DPM3|EARS2|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ECSIT|EIF2AK3|ELOVL4|ETFA|ETFB|ETFDH|ETHE1|FA2H|FARS2|FASTKD2|FECH|FGF14|FH|FOXG1|FOXRED1|FXN|GAA|GAD1|GAMT|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLA|GLDC|GLRX5|GLUD1|GNPAT|GPD2|GPHN|HADH|HADHA|HADHB|HARS2|HAX1|HCCS|HFE|HK1|HLCS|HMGCL|HMGCS2|HSD17B10|HSD3B2|HSPD1|IARS2|IDH2|ISCU|ITPR1|IVD|KARS|KCNC3|KCNJ11|KIAA0196|KIAA0226|KIF1B|KIF5A|LARS2|LETM1|LMBRD1|LRPPRC|MAOA|MAPT|MARS2|MCCC1|MCCC2|ME2|MECP2|MFN2|MFSD8|MGAT2|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MPV17|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|MYH7|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA3|NDUFA4|NDUFA5|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAB1|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB1|NDUFB10|NDUFB11|NDUFB2|NDUFB3|NDUFB4|NDUFB5|NDUFB6|NDUFB7|NDUFB8|NDUFB9|NDUFC1|NDUFC2|NDUFS					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NEFL|NIPA1|NKX2-1|NPC1|NPC2|NUBPL|OAT|OGDH|OPA1|OPA3|OTC|OXA1L|OXCT1|PAFAH1B1|PANK2|PARL|PARS2|PC|PCCA|PCCB|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX13|PHB|PHYH|PLP1|PMM2|PNKD|PNPLA2|PNPLA3|POLG|POLG2|PPM1B|PPOX|PPT1|PREPL|PRKCG|PRODH|PUS1|PWAR1|RARS2|REEP1|RFT1|RMRP|RNASEH2A|RNASEH2B|RNASEH2C|RRM2B|RYR1|SAMHD1|SARS2|SCN1A|SCO1|SCO2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SGCD|SLC12A3|SLC16A2|SLC19A2|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A16|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC29A3|SLC2A1|SLC2A10|SLC33A1|SLC35A1|SLC35C1|SLC3A1|SLC52A1|SLC6A8|SLC7A9|SOD1|SPAST|SPG11|SPG20|SPG7|SPTBN2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TACO1|TARS2|TAZ|TIMM8A|TK2|TMEM126A|TMEM70|TOP1MT|TPM2|TPP1|TRMU|TSFM|TTBK2|TUFM|TYMP|UBE3					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		A|UCP1|UCP2|UCP3|UNG|UQCR10|UQCR11|UQCRB|UQCRC1|UQCRC2|UQCRFS1|UQCRH|UQCRQ|UROS|USMG5|VARS2|WARS2|WFS1|XDH|XPNPEP3|YARS2|ZFYVE26					
GTR000509336.2	Transgenomic Transgenomic Connecticut 07D0995237 509336	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		FGFR3|FH|FKRP|FKTN|FLNA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|P					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		EX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000205580.1	United States Air Force, DNA	MECP2	Sequence analysis of the	Bi-directional Sanger	Peripheral (whole)	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostic Laboratory United States Air Force Mississippi NR 205580		entire coding region	Sequence Analysis	blood	answer	

NR: Not reported

Table D-8. Genetic tests for Rubinstein-Taybi syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511929.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511929	CREBBP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Multiplex Ligation-dependent Probe Amplification (MLPA |Bi-directional Sanger Sequence Analysis	—	—	No
GTR000511930.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511930	CREBBP	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	—	—	No
GTR000511931.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511931	CREBBP	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	—	—	No
GTR000511932.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511932	CREBBP	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	—	—	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000511933.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511933	CREBBP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	—	—	No
GTR000511934.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 511934	CREBBP	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	—	—	No
GTR000320329.1	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 320329	16p13.3	FISH-metaphase	Other	Amniocytes|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole)	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Massachusetts 22D2035168 514882	1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2			blood|Plasma|Saliva|Serum|Skin		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000502732.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502732	CREBBP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	—	No	No
GTR000502734.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502734	CREBBP	Deletion/duplication analysis	Comparative Genomic Hybridization	—	No	No
GTR000502735.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502735	EP300	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	—	No	No
GTR000512423.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512423	ATRX|BLM|BTK|CREBBP|CUL7|DHCR7|EP300|ERCC6|ERCC8|FGD1|GH1|GHR|GHRHR|GLI2|HESX1|IGF1|IGF1R|INSR|KDM6A|KMT2D|KRAS|LHX3|NBN|NIPBL|PITX2|POU1F1|PROP1|PTPN11|RAF1|ROR2|RPS6KA3|SHOX|SHOX2|SMARCAL1|SMC1A|SMC3|SOS1|SOX2|SOX3|SRCAP|STA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	—	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		T5B|TBCE|THRB|TRIM37|WRN					
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	—	No	No
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6					
GTR000510913.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510913	CREBBP|DHCR7|EP300|FGD1|KRAS|NIPBL|PTPN11|RAF1|SMC1A|SOS1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000320336.2	Genetic Services Laboratory University of Chicago Illinois 14D0917593 320336	CREBBP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000330884.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593	EP300	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Frozen	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	330884				tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva		
GTR000500145.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500145	CREBBP	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000500163.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 500163	EP300	Deletion/duplication analysis	Multiplex Ligation-dependent Probe Amplification (MLPA)	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Peripheral (whole) blood|Product of conception (POC)|Saliva	Decline to answer	Yes
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|H	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	28D0454363 327662	PRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2					
GTR000503218.3	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503218	CREBBP	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000503219.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503219	CREBBP	Deletion/duplication analysis	Microarray	—	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000503220.3	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503220	EP300	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Buccal swab|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000503221.2	Molecular Genetics Laboratory Children's Hospital Colorado Colorado 06D0644348 503221	EP300	Deletion/duplication analysis	Microarray	—	Decline to answer	No

Table D-9. Genetic tests for Smith-Magenis syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512797.2	Athena Diagnostics Inc Massachusetts 22D0069726 512797	ATP2A2|ATP6V0A2|CCDC88C|CLCNKA|CLCNKB|KCNA1|KCNJ1|KCNJ10|KIAA1279|KMT2D|LBR|LGI1|NIPBL|PANK2|PIGV|PLA2G6|RAI1|SERPINI1|SETBP1|SMC3|SYNGAP1|TBX1|TSC1|TSC2|VPS13A|VPS13B	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes
GTR000512796.2	Athena Diagnostics Inc Massachusetts 22D0069726 512796	ALDH7A1|ARFGEF2|ARHGEF9|ARX|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATRX|CACNA1A|CASK|CASR|CCDC88C|CDKL5|CHRNA2|CHRNA4|CHRNB2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COL18A1|COL4A1|CPT2|CSTB|CTSD|CUL4B|DCX|DEPDC5|DNAJC5|EFHC1|EMX2|EPM2A|FGD1|FGFR3|FKRP|FKTN|FLNA|FOXG1|GABRA1|GABRB3|GABRD|GABRG2|GPC3|GPR56|GRIA3|GRIN2A|HSD17B10|KCNA1|KCNJ1|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|LAMA2|LARGE|LBR|LGI1|MBD5|ME2|MECP2|MEF2C|MFSD8|NHLRC1|NIPBL|NOTCH3|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PCDH19|PEX7|PHF6|PIGV|PLA2G6|PLP1|PNKP|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRRT2|RAB39B|RAB3GAP	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|RAI1|RELN|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SERPINI1|SETBP1|SLC25A22|SLC2A1|SLC4A10|SLC9A6|SMC1A|SMC3|SMS|SNAP29|SPTAN1|SRPX2|STXBP1|SYNGAP1|SYP|TBC1D24|TBX1|TCF4|TPP1|TREX1|TSC1|TSC2|TUBA1A|TUBA8|TUBB2B|UBE3A|VPS13A|VPS13B|WDR62|ZEB2					
GTR000512125.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512125	RAI1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512124.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 512124	RAI1	Targeted variant analysis	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No
GTR000512123.1	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas	RAI1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	45D0660090 512123						
GTR000019599.1	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 19599	17p11.2	FISH-metaphase	Other	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Cystic hygroma fluid|Fetal blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000206875.1	Center for Human Genetics, Inc Massachusetts 22D0650242 206875	17p11.2	FISH-metaphase	Fluorescence in situ Hybridization (FISH)	Not reported	Decline to answer	No
GTR000295249.1	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 295249	17p11.2	FISH-metaphase	Metaphase FISH analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000514882.1	Courtagen Diagnostics	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel	Buccal swab|Cord blood|Fresh	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2		sequencing (MPS)	tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin		

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500883	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|BRD2|BTD|BUB1B|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FG	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		FR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDH2|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000325583.1	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center Ohio 36D0965797 325583	17p11.2	FISH-metaphase	FISH	Peripheral (whole) blood	Decline to answer	Yes
GTR000509083.1	Cytogenetics and Molecular Pathology Laboratory DLP Marquette General Hospital, LLC Michigan 23D1062707 509083	RAI1	FISH-interphase|FISH-metaphase|Karyotyping	FISH|FISH|G-banding	Buccal swab|Cord blood|Fetal blood|Peripheral (whole) blood	No	Yes
GTR000500042.2	Cytogenetics Laboratory ARUP Laboratories, Inc. Utah 46D0523979 500042	15q11.2-q13|17p11.2|17p13.3|22q11.2|22q13.3|4p16.3|5p15.2|7q11.23|Xp22.3|Yp11.3	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000013536.1	Cytogenetics Laboratory Indiana University School of Medicine Indiana 15D0647198 13536	17p11.2	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization (FISH)|Fluorescence in situ hybridization (FISH)	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin|Urine	Decline to answer	Yes
GTR000512587.2	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512587	ADSL|AFF2|AP1S2|ARX|ATRX|BCKDK|BRAF|CACNA1C|CASK|CDKL5|CHD7|CNTNAP2|CREBBP|DHCR7|DMD|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|HPRT1|KDM5C|L1CAM|MAGEL2|MBD5|MECP2|MED12|MEF2C|MID1|NHS|NIPBL|NLGN3|NLGN4X|NR1I3|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RELN|SCN1A|SLC2A1|SLC9A6|SMARCB1|SMC1A|TCF4|UBE2A|UBE3A|VPS13B|ZEB2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No
GTR000512586.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512586	ABCD1|ACSL4|AFF2|AP1S2|ARHGEF9|ARX|ATP6AP2|ATP7A|ATRX|BCOR|BRWD3|CASK|CCDC22|CDK16|CDKL5|CLIC2|CNKSR2|CUL4B|DCX|DKC1|DLG3|DMD|FANCB|FGD1|FLNA|FMR1|FRMPD4|FTSJ1|GDI1|GK|GPC3|GRIA3|HCCS|H	Deletion/duplication analysis|Sequence analysis of the entire coding region	Microarray|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Not reported	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		CFC1|HPRT1|HSD17B10|HUWE1|IDS|IGBP1|IL1RAPL1|IQSEC2|KDM5C|KIAA2022|KLF8|L1CAM|LAMP2|MAOA|MBTPS2|MECP2|MED12|MID1|NAA10|NDP|NDUFA1|NHS|NLGN3|NLGN4X|NSDHL|OCRL|OFD1|OPHN1|OTC|PAK3|PCDH19|PDHA1|PGK1|PHF6|PHF8|PLP1|PORCN|PQBP1|PRPS1|PTCHD1|RAB39B|RBM10|RPL10|RPS6KA3|SHROOM4|SLC16A2|SLC9A6|SMC1A|SMS|SOX3|SYN1|SYP|TIMM8A|TSPAN7|UBE2A|UPF3B|ZDHHC15|ZDHHC9|ZNF711					
GTR000502986.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502986	RAI1	Deletion/duplication analysis	Comparative Genomic Hybridization	Not reported	No	No
GTR000502985.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 502985	RAI1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Not reported	No	No
GTR000509442.11	Fulgent Clinical Diagnostics Lab	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|	Deletion/duplication analysis|Sequence analysis	Next-Generation (NGS)/Massively parallel	Not reported	Not reported	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Fulgent Diagnostics California 05D2043189 509442	ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRI	of the entire coding region	sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)			

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		N2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|MANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509436.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509436	ABCC8|ACY1|ADAMTSL2|ADSL|AGA|ALDH4A1|ALDH5A1|ALDH7A1|AMT|ANTXR2|ARG1|ARSA|ARSB|ASAH1|ASPA|ATP13A2|BTD|CLN3|CLN5|CLN6|CLN8|COL11A2|COL2A1|CTNS|CTSA|CTSC|CTSD|CTSK|DHCR7|DNAJC5|DPYD|DYM|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ETFA|ETFB|ETFDH|FH|FOLR1|FUCA1|GAA|GALC|GALNS|GAMT|GBA|GCDH|GCSH|GLA|GLB1|GLDC|GM2A|GNE|GNPTAB|GNPTG|GNS|GPC3|GUSB|HEXA|HEXB|HGSNAT|HPD|HRAS|IDS|IDUA|L2HGDH|LAMA2|LAMP2|LIPA|LMBRD1|MAN2B1|MANBA|MCOLN1|MFSD8|MOCS1|MOCS2|NAGA|NAGLU|NEU1|NPC1|NPC2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX26|PEX3|PEX5|PEX6|PGK1|PHYH|PPT1|PRODH|PSAP|QDPR|RAI1|SGSH|SLC17A5|SLC25A15|SLC46A1|SMPD1|SUMF1|SUOX|TCF4|TPP1					
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509399	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000509342.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509342	ANKRD11|AP1S2|ARX|ATRX|AUTS2|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CDKL5|CHD7|CHD8|CNTNAP2|CNTNAP5|CREBBP|DHCR7|DLGAP2|DMD|DOCK4|DPP10|DPP6|EHMT1|FGD1|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GABRG1|GNA14|GRIN2B|GRPR|HOXA1|HPRT1|IMMP2L|KATNAL2|KCTD13|KDM5C|KIRREL3|KLHL3|L1CAM|LAMC3|MBD5|MECP2|MED12|MEF2C|MET|MID1|NEGR1|NHS|NIPBL|NLGN3|NLGN4X|NRXN1|NSD1|NTNG1|OPHN1|PAFAH1B1|PCDH19|PCDH9|PDE10A|PHF6|PIP5K1B|PNKP|PON3|PQBP1|PTCHD1|PTEN|PTPN11|RAB39B|RAI1|RBFOX1|RELN|RPL10|SATB2|SCN1A|SCN2A|SHANK2|SHANK3|SLC6A4|SLC9A6|SLC9A9|SMC1A|SMG6|SNRPN|SOX5|SPAST|ST7|STK3|TCF4|TSC1|TSC2|UBE3A|VPS13B|ZEB2|ZNF507|ZNF804A|ZNHIT6	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cell culture|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000327662.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska	AP1S2|ARX|ASPM|ATRX|AVPR1A|BDNF|BRAF|CACNA1C|CASK|CBL|CDKL5|CHD7|CNTNAP2|CREBBP|DCX|DHCR7|DMD|EHMT1|ERCC6|ERCC8|FGD1|FGFR1|FGFR2|FGFR3|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|HDAC8|HOXA1|H	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	28D0454363 327662	PRT1|HRAS|KDM5C|KMT2D|KRAS|L1CAM|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MEF2C|MET|MID1|MKKS|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|OPHN1|PAFAH1B1|PCDH19|PHF6|PNKP|PQBP1|PTCH1|PTEN|PTPN11|RAB39B|RAD21|RAF1|RAI1|RELN|RPGRIP1L|RPS6KA3|SCN1A|SHANK2|SHANK3|SHOC2|SLC2A1|SLC6A4|SLC9A6|SMC1A|SMC3|SOS1|SPRED1|TCF4|TSC1|TSC2|TUBA1A|UBE3A|VPS13B|ZEB2					
GTR000500650.2	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500650	17q11.2	FISH-metaphase	Other	Amniocytes|Bone marrow|Cell culture|Chorionic villi|Cord blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin	No	Yes
GTR000507774.1	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 507774	RAI1	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Bi-directional Sanger Sequence Analysis	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole)	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
					blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood cell prep		
GTR000504366.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504366	17p11.2	FISH-metaphase	Other	Peripheral (whole) blood	No	Yes
GTR000506048.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506048	17p11.2	FISH-metaphase	Other	Amniocytes|Peripheral (whole) blood	No	Yes
GTR000509336.2	Transgenomic Transgenomic Connecticut 07D0995237 509336	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|C	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SM					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		C1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000305783.1	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305783	17p11.2	FISH-interphase|FISH-metaphase|Deletion/duplication analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray	Amniocytes|Amniotic fluid|Cord blood|Fibroblasts|Peripheral (whole) blood	Decline to answer	Yes

Table D-10. Genetic tests for Velocardiofacial syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000512796.2	Athena Diagnostics Inc Massachusetts 22D0069726 512796	ALDH7A1|ARFGEF2|ARHGEF9|ARX|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATRX|CACNA1A|CASK|CASR|CCDC88C|CDKL5|CHRNA2|CHRNA4|CHRNB2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COL18A1|COL4A1|CPT2|CSTB|CTSD|CUL4B|DCX|DEPDC5|DNAJC5|EFHC1|EMX2|EPM2A|FGD1|FGFR3|FKRP|FKTN|FLNA|FOXG1|GABRA1|GABRB3|GABRD|GABRG2|GPC3|GPR56|GRIA3|GRIN2A|HSD17B10|KCNA1|KCNJ1|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|LAMA2|LARGE|LBR|LGI1|MBD5|ME2|MECP2|MEF2C|MFSD8|NHLRC1|NIPBL|NOTCH3|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PCDH19|PEX7|PHF6|PIGV|PLA2G6|PLP1|PNKP|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRRT2|RAB39B|RAB3GAP1|RAI1|RELN|RNASEH2A|RNASEH2B|RNASEH2C|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SERPINI1|SETBP1|SLC25A22|SLC2A1|SLC4A10|SLC9A6|SMC1A|SMC3|SMS|SNAP29|SPTAN1|SRPX2|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		STXBP1|SYNGAP1|SYP|TBC1D24|TBX1|TCF4|TPP1|TREX1|TSC1|TSC2|TUBA1A|TUBA8|TUBB2B|UBE3A|VPS13A|VPS13B|WDR62|ZEB2					
GTR000512797.2	Athena Diagnostics Inc NR Massachusetts 22D0069726 512797	ATP2A2|ATP6V0A2|CCDC88C|CLCNKA|CLCNKB|KCNA1|KCNJ1|KCNJ10|KIAA1279|KMT2D|LBR|LGI1|NIPBL|PANK2|PIGV|PLA2G6|RAI1|SERPINI1|SETBP1|SMC3|SYNGAP1|TBX1|TSC1|TSC2|VPS13A|VPS13B	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	Yes
GTR000335344.1	BayCare Cytogenetics Laboratory BayCare Laboratories, LLC Florida 10D2029560 335344	HIRA	FISH-metaphase	Digital / Virtual karyotyping	Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000508280.2	Baylor Medical Genetics Laboratories Baylor College of Medicine Texas 45D0660090 508280	AARS2|AASS|ABAT|ABCA12|ABCA4|ABCB11|ABCB4|ABCB6|ABCB7|ABCD1|ABHD12|ABHD5|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACO2|ACOX1|ACSF3|ACSL4|ADAM9|ADCK3|ADSL|AFG3L2|AGK|AGL|AGPS|AGXT|AIFM1|AIPL1|AK1|AK2|AKAP10|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|ALDOA|AL	Deletion/duplication analysis	Comparative Genomic Hybridization	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		DOB|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|ALMS1|AMACR|AMER1|AMN|AMT|ANKH|ANKRD26|AP3B1|APP|APTX|ARG1|ARL6|ASL|ASS1|ATIC|ATP5E|ATP6V0A2|ATP7A|ATP7B|ATP8B1|ATPAF2|AUH|B4GALT1|B4GALT7|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|BCKDHA|BCKDHB|BCOR|BCS1L|BEST1|BLOC1S3|BOLA3|BRCA1|BTD|C10orf2|C12orf65|C1QTNF5|C2orf71|C8orf37|CA2|CA4|CABP4|CACNA1F|CACNA2D4|CASP8|CAT|CBS|CC2D2A|CCDC28B|CCDC39|CDH23|CDHR1|CEP290|CERKL|CHAT|CHM|CISD2|CLCN7|CLN3|CLRN1|CNGA1|CNGA3|CNGB1|CNGB3|COG1|COG7|COG8|COL1A1|COL1A2|COL2A1|COL3A1|COL5A1|COL5A2|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|CPOX|CPS1|CPT1A|CPT2|CRB1|CRTAP|CRX|CRYAB|CTSA|CTSD|CTSK|CUBN|CYB5A|CYB5R3|CYBA|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP17A1|CYP1B1|CYP24A1|CYP27A1|CYP27B1|CYP4V2|D2HGDH|DARS2|DBT|DDOST|DFNB31|DGUOK|DHDDS|DHODH|DIABLO|DLAT|DLD|DMGDH|DNAJC19|DNM1L|DPM1|DPM3|DSP|DTN					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		BP1|EFEMP1|ELAC2|ELN|ELOVL4|ENO3|ETFA|ETFB|ETFDH|ETHE1|EYA1|EYA4|EYS|FAH|FAM161A|FAM20C|FASTKD2|FBLN5|FBN1|FBP1|FECH|FH|FKBP10|FLVCR1|FOXC1|FOXL2|FOXRED1|FRMD7|FSCN2|FXN|FYCO1|FZD4|G6PC|GAA|GAD1|GALC|GALE|GALK1|GALNS|GALT|GAMT|GARS|GATM|GBE1|GCDH|GCK|GCKR|GCSH|GFER|GFM1|GIF|GJA3|GK|GLB1|GLDC|GLRX5|GLUD1|GLYCTK|GM2A|GNAT1|GNAT2|GNE|GNPTAB|GNS|GOT1|GPD1|GPD2|GPI|GPR143|GPR98|GPX1|GRK1|GRM6|GRN|GSN|GUCA1A|GUCA1B|GUCY2D|GUSB|GYS1|GYS2|HADHA|HADHB|HAGH|HARS|HAX1|HBB|HCCS|HESX1|HEXA|HEXB|HGSNAT|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HNF1A|HNF1B|HP|HPRT1|HPS1|HPS3|HPS4|HPS5|HPS6|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH2|IDH3B|IMPDH1|IMPG2|INPP5E|INVS|IQCB1|ISCU|IVD|JAG1|KARS|KCNJ13|KCNV2|KIF1B|KIF21A|KLHL7|KRT12|KRT3|KRT5|L2HGDH|LCA5|LDHA|LDHB|LEMD3|LEPRE1|LIAS|LMBRD1|LMX1B|LPIN1|LRAT|LRP5|LRPPRC|MAN2B1|MANBA|MAOA|MC1R|MCCC1|MCCC2|MC					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		EE|ME2|MECP2|MEF2A|MEN1|MERTK|MFN2|MFRP|MGAT2|MKKS|MKS1|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MPDU1|MPI|MPV17|MRPL3|MRPS16|MRPS22|MSRB3|MTHFR|MTO1|MTPAP|MTR|MTRR|MUT|MUTYH|MYO7A|MYOC|NAGLU|NAGS|NCOA4|NDP|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFAF5|NDUFAF6|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NEFH|NEUROD1|NFU1|NHS|NME1|NPHP1|NPHP3|NPHP4|NRL|NT5C3A|NUBPL|NYX|OAT|OCA2|OCRL|OGDH|OGG1|OPA1|OPA3|OPN1LW|OPN1MW|OPTN|OSTM1|OTC|OTX2|OXCT1|PAH|PANK2|PARK2|PARK7|PAX2|PAX6|PC|PCCA|PCCB|PCDH15|PCK2|PDE6A|PDE6B|PDE6C|PDE6G|PDHA1|PDHB|PDP1|PDSS1|PDSS2|PDZD7|PFKM|PGAM2|PGM1|PHB|PHKA1|PHKA2|PHKB|PHKG2|PHOX2A|PHYH|PINK1|PITPNM3|PITX2|PITX3|PLA2G2A|PLOD2|PLOD3|PMM2|PNKD|PNPLA2|POLG|POLG2|PPARG|PPARGC1B|PPIB|PPOX|PPP2R1B|PRCD|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		PRKCG|PRODH|PROM1|PRPF31|PRPF6|PRPF8|PRPH2|PSAP|PSEN1|PTRF|PTS|PUS1|PYCR1|PYGL|PYGM|QDPR|RAF1|RARS2|RAX|RB1|RD3|RDH12|RDH5|REEP1|RET|RFT1|RGR|RGS9|RHO|RILP|RIMS1|RLBP1|RNASEL|RP1|RP1L1|RP2|RP9|RPE65|RPGR|RPGRIP1|RPL35A|RPS14|RRM2B|RS1|SAG|SARDH|SARS2|SCO2|SCP2|SDHAF1|SDHAF2|SDHB|SDHC|SECISBP2|SEMA4A|SEPT9|SERPINF1|SGSH|SHH|SIX6|SLC16A1|SLC22A4|SLC22A5|SLC24A1|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC26A4|SLC34A1|SLC35A1|SLC35C1|SLC37A4|SLC39A13|SLC3A1|SLC45A2|SLC9A3R1|SLC9A6|SMPD1|SNCB|SNRNP200|SOD1|SOD2|SOST|SOX2|SP7|SPATA7|SPG7|SPR|SPTLC2|SQSTM1|SRD5A3|STAR|STAT1|STAT3|STRA6|STXBP1|SUCLA2|SUCLG1|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TCN2|TEAD1|TGFB1|TGFBI|TIMM8A|TIMP3|TK2|TLR3|TLR4|TMEM126A|TMEM127|TMEM67|TMEM70|TNFRSF11A|TNFRSF11B|TNFSF11|TOPORS|TP53|TPP1|TRIM32|TRMU|TRPM1|TSFM|TSPAN12|T					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		TC19|TTC8|TUBA1A|TUBB3|TUFM|TULP1|TUSC3|TYMP|TYR|TYROBP|TYRP1|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|USH1C|USH1G|USH2A|VCP|VHL|VSX1|WFS1|WT1|WWOX|XDH|XPNPEP3|YARS2|ZEB1|ZFHX3|ZNF513					
GTR000257087.2	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 257087	22q11.2	FISH-metaphase	Other	Amniocytes|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000206879.1	Center for Human Genetics, Inc NR Massachusetts 22D0650242 206879	22q11.2	FISH-interphase|FISH-metaphase	Fluorescence in situ Hybridization (FISH)|Fluorescence in situ Hybridization (FISH)	NR	Decline to answer	No
GTR000295251.1	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical	22q11.2	FISH-metaphase	Metaphase FISH analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral (whole) blood|Product of	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Center Ohio 36D0656333 295251				conception (POC)		
GTR000500883.8	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500883	ABAT|ABCC2|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|AKT3|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|ALG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASAH1|ASPA|ASPM|ATIC|ATN1|ATP1A2|ATP1A3|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDHA|BCKDHB|BCKDK|BCS1L|BRAF|BRAT1|BRD2|BTD|BUB1B|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT1A|CPT2|CSTB|CTSA|CTSD|CTSF|CUL4B|CYP1B1|CYP2A6|CYP2B6|CYP2C19|CYP2C9|CYP2D6|CYP2R1|CYP2U1|CYP3A5|DBT|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FAAH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FOXH1|FUCA1|GABBR2|GABRA1|GABRA2|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDH2|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIAS|LIG4|LRPPRC|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED12|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NGLY1|NHEJ1|NHLR					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		C1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPA1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PIK3CA|PIK3R2|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|RTTN|SAMHD1|SCARB2|SCN10A|SCN11A|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGCE|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SNIP1|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|ST3GAL5|STIL|STRADA|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TB					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		C1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TRPM6|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000514882.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 514882	ADSL|AFF2|ANK3|AP1S2|AP4B1|AP4E1|AP4M1|AP4S1|ARFGEF2|ARX|ATRX|BBS4|BCKDK|BCOR|BDNF|BRAF|C12orf57|CA8|CACNA1C|CACNG2|CASK|CC2D1A|CDH15|CDKL5|CHD7|CLIC2|CNTNAP2|COMT|CRBN|CREBBP|CTCF|CTNNB1|D2HGDH|DCX|DDHD2|DHCR7|DMD|EHMT1|EP300|ERLIN2|FGD1|FGF8|FMR1|FOLR1|FOXG1|FOXP1|FOXP2|GABRB3|GATAD2B|GNS|GRIA3|GRIK2|GRIN1|GRIN2B|HCFC1|HDAC8|HGSNAT|HOXA1|HPRT1|HRAS|HTR2A|HYDIN|IDH2|KCNJ10|KDM5C|KIAA2022|KRAS|L1CAM|LINS|LRP2|MAN1B1|MAP2K1|MAP2K2|MBD5|MECP2|MED12|MED23|MEF2C|MET|MID1|MKKS|NAGLU|NF1|NHS|NIPBL|NLGN3|NLGN4X|NRAS|NRXN1|NSD1|NSDHL|NSUN2|OCRL|OPHN1|PAFAH1B1|PCDH19|PCNT|PHF6|PLP1|PNKP|PQBP1|PRSS12|PTEN|PTPN11|RAB39B|RAF1|RAI1|RELN|RPS6KA3|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Saliva|Serum|Skin	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		SCN1A|SGSH|SHANK2|SHANK3|SHOC2|SHROOM4|SLC25A1|SLC2A1|SLC6A8|SLC9A6|SLC9A9|SMARCB1|SMC1A|SMC3|SNRPN|SOBP|SOS1|SRPX2|TBX1|TCF4|TECR|TRAPPC9|TSC1|TSC2|TUSC3|UBE2A|UBE3A|UPF3B|VLDLR|VPS13B|ZEB2					
GTR000500619.2	Cytogenetics and Microarray Laboratory Kennedy Krieger Institute Maryland 21D0649789 500619	22q11.2|Yp11.3	Fluorescence in situ hybridization (FISH)	Other	Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood	Decline to answer	Yes
GTR000325584.2	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center Ohio 36D0965797 325584	22q11.2	FISH-interphase|FISH-metaphase	FISH|FISH	Amniocytes|Amniotic fluid|Fetal blood|Peripheral (whole) blood	Decline to answer	Yes
GTR000316013.2	Cytogenetics and Molecular Pathology Laboratory DLP Marquette General Hospital, LLC Michigan	HIRA	FISH-interphase|FISH-metaphase|Karyotyping	FISH LSI HIRA/SHANK3|FISH LSI HIRA/SHANK3|G-banding	Buccal swab|Cell culture|Cord blood|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	23D1062707 316013						
GTR000013538.2	Cytogenetics Laboratory Indiana University School of Medicine Indiana 15D0647198 13538	22q11.2	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization (FISH)|Fluorescence in situ hybridization (FISH)	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin|Urine	Decline to answer	Yes
GTR000032597.1	Cytogenetics Laboratory SUNY Upstate Medical University New York 33D0654590 32597	22q11.2	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin	Yes	Yes
GTR000500042.2	Cytogenetics Laboratory ARUP Laboratories, Inc. Utah 46D0523979 500042	15q11.2-q13|17p11.2|17p13.3|22q11.2|22q13.3|4p16.3|5p15.2|7q11.23|Xp22.3|Yp11.3	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501825.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501825	22q11.2	FISH-interphase	Fluorescence In Situ Hybridization	Fetal blood|Peripheral (whole) blood	Decline to answer	No
GTR000512325.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512325	ABCC9|ACTA2|ACTC1|ACTN2|ACVRL1|AKAP9|ANK2|ANKRD1|BAG3|BMPR2|BRAF|CACNA1C|CACNB2|CASQ2|CAV1|CAV3|CBS|COL3A1|COL5A1|COL5A2|CRYAB|CSRP3|DES|DMD|DSC2|DSG2|DSP|DTNA|EMD|ENG|FBN1|FBN2|FHL2|FKTN|FLNA|GATAD1|GLA|GPD1L|HCN4|HRAS|JPH2|JUP|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|KRAS|LAMA4|LAMP2|LDB3|LMNA|MAP2K1|MAP2K2|MED12|MYBPC3|MYH11|MYH6|MYH7|MYL2|MYL3|MYLK2|MYOZ2|MYPN|NEBL|NEXN|NKX2-5|NR2F2|NRAS|PKP2|PLN|PRKAG2|PTPN11|RAF1|RANGRF|RBM20|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SGCD|SKI|SLC2A10|SMAD3|SNTA1|SOS1|TAZ|TCAP|TGFB2|TGFBR1|TGFBR2|TMEM43|TMPO|TNNC1|TNNI3|TNNT2|TPM1|TTN|TTR|VCL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood	No	No
GTR000512328.1	Emory Genetics	AKAP9|ANK2|CACNA1C|C	Sequence analysis of the	Next-Generation	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Laboratory Emory University School of Medicine Georgia 11D0683478 512328	ACNB2|CASQ2|DSC2|DSG2|DSP|GPD1L|HCN4|JUP|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|NKX2-5|PKP2|RANGRF|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SNTA1|TMEM43	entire coding region	(NGS)/Massively parallel sequencing (MPS)			
GTR000512415.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512415	ACVR2B|AHI1|AIPL1|ARL13B|ARL6|ATXN10|B9D1|B9D2|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|C2orf71|C5orf42|CC2D2A|CCDC28B|CCDC39|CCDC40|CDH23|CEP164|CEP290|CEP41|CFTR|CLRN1|CRB1|CRELD1|CRX|DFNB31|DNAAF1|DNAAF2|DNAAF3|DNAH11|DNAH5|DNAI1|DNAI2|DNAL1|DYNC2H1|EVC|EVC2|FOXH1|GDF1|GLIS2|GPR98|GUCY2D|HYLS1|IFT43|IFT80|IMPDH1|INVS|IQCB1|KCNJ13|KIF7|LCA5|LEFTY2|LRAT|MKKS|MKS1|MYO7A|NEK1|NEK8|NKX2-5|NME8|NODAL|NPHP1|NPHP3|NPHP4|OFD1|PCDH15|PKD2|PKHD1|RD3|RDH12|RPE65|RPGR|RPGRIP1|RPGRIP1L|RSPH4A|RSPH9|SCNN1A|SCNN1B|SCNN1G|SDCCAG8|SPATA7|TCTN1|TCTN2|TMEM138|TMEM216|TMEM231|TMEM237|TMEM67|TOPORS|TRIM32|TSC1|TSC2|TTC21B|TTC8|TULP1|UMOD|USH1C	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|USH1G|USH2A|VHL|WDPCP|WDR19|WDR35|XPNPEP3|ZIC3|ZNF423					
GTR000512588.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512588	ACTB|ACTG1|AHI1|ARFGEF2|ARX|CASK|CC2D2A|CEP290|CEP41|CHMP1A|DCX|EOMES|EXOSC3|FKRP|FKTN|FLNA|GPR56|KIAA1279|KIF7|LAMC3|LARGE|MKS1|NPHP1|OPHN1|PAFAH1B1|POMGNT1|POMT1|POMT2|PQBP1|RAB3GAP1|RAB3GAP2|RARS2|RELN|RPGRIP1L|RTTN|SRPX2|TMEM138|TMEM216|TMEM237|TMEM67|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|VRK1|WDR62	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No
GTR000512596.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 512596	ABCC8|AGPAT2|AKT2|BLK|BMP15|BSCL2|CHD7|CIDEC|CISD2|CYP17A1|CYP19A1|EIF2AK3|FGF8|FGFR1|FIGLA|FOXP3|FSHR|GATA6|GCK|GDF9|GLIS3|GNRH1|GNRHR|HADH|HNF1A|HNF1B|HNF4A|IER3IP1|INS|INSR|KCNJ11|KISS1|KISS1R|KLF11|LHCGR|LMNA|NEUROD1|NOBOX|NR0B1|NR5A1|NSMF|PAX4|PDX1|POR|PPARG|PROK2|PROKR2|PSMC3IP|PTF1A|PTRF|RFX6|SLC2A2|TAC3|TACR3|TBC1D4|WFS1|ZMPSTE24	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No
GTR000512608.1	Emory Genetics Laboratory	ACTB|ACTG1|ADSL|AHI1|ALDH7A1|ARFGEF2|ARHGEF9|ARX|ASPM|ATP1A2|	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	No	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Emory University School of Medicine Georgia 11D0683478 512608	ATP6AP2|ATR|ATRX|BCKDK|CACNB4|CASC5|CASK|CC2D2A|CDC6|CDK5RAP2|CDKL5|CDT1|CENPJ|CEP135|CEP152|CEP290|CEP41|CEP63|CHMP1A|CHRNA2|CHRNA4|CHRNB2|CLN3|CLN5|CLN6|CLN8|CNTNAP2|CPA6|CSTB|CTSD|DCX|DHCR7|DISP1|DNAJC5|EFHC1|EHMT1|EOMES|EPM2A|EXOSC3|FGF8|FKRP|FKTN|FLNA|FOLR1|FOXG1|FOXH1|GABRA1|GABRG2|GAMT|GATM|GLI2|GOSR2|GPR56|GPR98|GRIN2A|GRIN2B|KCNJ10|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KIAA1279|KIF7|LAMC3|LARGE|LARP7|LGI1|LIAS|MAGI2|MAPK10|MBD5|MCPH1|MECP2|MEF2C|MFSD8|MKS1|MYCN|NDE1|NHLRC1|NIN|NODAL|NPHP1|NRXN1|OPHN1|ORC4|ORC6|PAFAH1B1|PCDH19|PCNT|PLCB1|PNKP|PNPO|POC1A|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRRT2|PTCH1|RAB18|RAB3GAP1|RAB3GAP2|RARS2|RBBP8|RELN|RPGRIP1L|RTTN|SCARB2|SCN1A|SCN1B|SCN2A|SCN8A|SCN9A|SHH|SIX3|SLC19A3|SLC25A19|SLC25A22|SLC2A1|SLC9A6|SPTAN1|SRPX2|ST3GAL3|ST3GAL5|STIL|STXBP1|SYN1|TBC1D24|TCF4|TGIF					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|TMEM138|TMEM216|TMEM237|TMEM67|TPP1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUBB3|UBE3A|VLDLR|VRK1|WDR62|ZEB2|ZIC2|ZNF335					
GTR000506437.10	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 506437	ABCC9|ACTC1|ACTN2|AKAP9|ANK2|ANKRD1|BAG3|CACNA1C|CACNA2D1|CACNB2|CALR3|CASQ2|CAV3|COX15|CRYAB|CSRP3|DES|DMD|DOLK|DSC2|DSG2|DSP|DTNA|EMD|EYA4|FKTN|FLNA|FXN|GAA|GATA4|GATAD1|GJA5|GLA|GPD1L|HCN4|ILK|JAG1|JPH2|JUP|KCNA5|KCNE1|KCNE2|KCNE3|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|LAMA4|LAMP2|LDB3|LMNA|MRPL3|MURC|MYBPC3|MYH6|MYH7|MYL2|MYL3|MYLK2|MYOM1|MYOZ2|MYPN|NDUFAF1|NEBL|NEXN|NKX2-5|NPPA|PDLIM3|PKP2|PLN|PRKAG2|PSEN2|PTPN11|RAF1|RBM20|RYR2|SCN1B|SCN3B|SCN4B|SCN5A|SDHA|SGCD|SNTA1|SYNE1|TAZ|TBX1|TBX5|TCAP|TGFB3|TMEM43|TMPO|TNNC1|TNNI3|TNNT2|TPM1|TTN|TTR|TXNRD2|VCL	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509335.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics	ABCC9|GATA4|GATA6|GJA5|HCN4|KCNA5|KCND3|KCNE1|KCNE2|KCNH2|KCNJ2|KCNJ5|KCNJ8|KCNQ1|LMNA|NPPA|NUP155|RYR2|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	California 05D2043189 509335	SCN1B|SCN2B|SCN3B|SCN4B|SCN5A					
GTR000509399.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509399	ABAT|ABCB1|ABCC8|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG12|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|APTX|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP6AP2|ATP6V0A2|ATPAF2|ATR|ATRX|B4GALT1|BCS1L|BRAF|BTD|BUB1B|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCL2|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLCNKA|CLCNKB|CLN3|CLN5|CLN6|CLN8|CNTNAP2|COG1|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|EFHC1|EFHC2|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FLVCR2|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GNE|	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		GNPTAB|GNPTG|GNS|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|GUSB|HCN1|HCN4|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|IDS|IDUA|KAT6B|KCNA1|KCNJ1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNV2|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LIG4|LRPPRC|MAGI2|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|MECP2|MED17|MEF2C|MFSD8|MGAT2|MLC1|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHEJ1|NHLRC1|NIPBL|NODAL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDSS1|PDSS2|PEX1|PEX12|PEX14|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PHF6|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RELN|RFT1|RNASEH2A|RNASEH2B|RN					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN10A|SCN1A|SCN1B|SCN2A|SCN2B|SCN3A|SCN3B|SCN4A|SCN4B|SCN5A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC17A5|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35C1|SLC46A1|SLC6A5|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRPX2|STIL|STXBP1|SUMF1|SUOX|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|UBE3A|VANGL1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000509442.11	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509442	ABCC6|ABCD1|ABCG5|ACAT1|ACOX1|ACSL4|ACY1|ADAR|ADSL|AFF2|AFP|AGL|AGT|AGTR2|AHI1|AIFM1|ALDH18A1|ALDH4A1|ALG11|ALG12|ALG6|ALX4|AMER1|AP1S1|AP1S2|AP3B1|AP4B1|AP4E1|AP4M1|AP4S1|APOB|AQP7|AR|ARG1|ARHGEF6|ARID1A|ARID1B|ARX|ASPM|ASS1|ATL1|ATM|ATP13A2|ATP1A2|ATP6AP2|ATP7A|ATRX|AUH|AVP|AVPR2|BBS9|BCS1L|BIN1|BRCA2|BRIP1|BRWD3|BUB1B|CACNG2|CAMTA	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|CANT1|CASK|CBS|CC2D1A|CC2D2A|CCDC88C|CDH15|CDKL5|CDKN1C|CEP290|CEP41|CEP57|CHD7|CHRNA4|CLN3|CNTNAP2|COG5|COG7|COL1A2|CP|CPA6|CPS1|CRADD|CRBN|CTC1|CTNNB1|CTSA|CUL4B|CYB5R3|CYP27A1|D2HGDH|DARS2|DBT|DHCR24|DHCR7|DIP2B|DLG3|DMD|DPYD|DYNC1H1|DYRK1A|EBP|EFNB1|EHMT1|ELOVL4|ERCC2|ERCC3|ERCC5|ERCC6|ERCC8|F5|FAM126A|FANCG|FBLN5|FBN1|FBN2|FGD1|FGF14|FGFR1|FGFR2|FGFR3|FKRP|FKTN|FMR1|FOXG1|FOXP1|FTO|FTSJ1|G6PC3|GABRG2|GALE|GAMT|GAN|GBA|GBE1|GCK|GDI1|GFAP|GFM1|GHR|GLI3|GLRA1|GLUL|GLYCTK|GM2A|GNAS|GNPAT|GNPTAB|GNPTG|GRIA3|GRIK2|GRIN1|GRIN2A|GRIN2B|GRM1|GSS|GUSB|GYS2|HAX1|HDAC4|HDAC8|HEPACAM|HEXB|HOXD10|HPD|HSD17B10|HSPD1|HUWE1|IDS|IGBP1|IGF1|IGF1R|IL1RAPL1|INSR|IQSEC2|IRX5|ITGA7|KCNJ10|KCNJ11|KCNK9|KCNQ2|KCTD7|KDM5C|KIF11|KIF1A|KIF21A|KIF5A|KIF7|KIRREL3|KRAS|L1CAM|LAMA2|LAMP2|LARGE|LBR|LHX3|LIG4|LMBRD1|LRP5|LYST|MAGT1|MAN1B1|MAN2B1|M					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		ANBA|MAPT|MAT1A|MBD5|MBTPS2|MCCC1|MCCC2|MCOLN1|MCPH1|MECP2|MED17|MED23|MEF2C|MFSD8|MGAT2|MKKS|MMADHC|MOCS2|MPI|MPZ|MRAP|MTFMT|MTHFR|MTR|MYCN|MYO5A|MYO7A|NAGA|NBN|NDP|NDUFA1|NDUFAF5|NDUFS1|NF1|NGF|NHEJ1|NHP2|NIPBL|NLGN3|NPC1|NPC2|NPHP3|NRXN1|NSDHL|NSUN2|OFD1|OPHN1|ORC1|PAFAH1B1|PAH|PAK3|PAX6|PCDH19|PCNT|PDE4D|PDHX|PDSS1|PEX7|PGK1|PHF8|PHKA2|PHKG2|PIGL|PIGO|PIGV|PLA2G6|PLP1|POMGNT1|POMT1|POMT2|POU1F1|PPOX|PQBP1|PRICKLE1|PRKAR1A|PRSS12|PTEN|PTPN11|PYCR1|PYGL|RAB39B|RAB40AL|RAI1|RAPSN|RBBP8|RBM10|RFX6|RPGRIP1L|RPS6KA3|SACS|SAMHD1|SATB2|SCN1A|SCN8A|SDCCAG8|SGCA|SGSH|SHANK2|SHANK3|SHROOM4|SIL1|SLC16A2|SLC20A2|SLC25A12|SLC25A13|SLC25A15|SLC2A1|SLC2A2|SLC35C1|SLC46A1|SLC4A4|SLC5A2|SLC5A5|SLC6A4|SLC6A8|SLC7A7|SLC9A6|SLX4|SMARCA4|SMARCB1|SMC1A|SMS|SNIP1|SOBP|SOX10|SOX2|SOX3|SPR|SPTAN1|SPTLC1|SRD5A3|SRPX2|ST3GAL3|STAT5B					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		|STRA6|STX11|STXBP1|SUCLG1|SYNGAP1|SYP|SYT14|TBC1D24|TBCE|TBX1|TECR|TGIF1|TH|THRB|TINF2|TMCO1|TMEM165|TMEM216|TMEM67|TMEM70|TPH2|TPK1|TRAPPC9|TRHR|TSC1|TSC2|TSHR|TSPAN7|TTC37|TTR|TUBA1A|TUBA8|TUBB2B|TUBB3|TUSC3|TWIST1|UBE2A|UBE3A|UPB1|UPF3B|UROC1|USP9X|VLDLR|VPS13B|WDR62|WDR81|WRN|XIST|XPNPEP3|ZBTB16|ZBTB24|ZDHHC9|ZEB2|ZFP57|ZFYVE26|ZIC2					
GTR000509445.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509445	ABCC8|AKT2|BLK|CEL|CISD2|CP|EIF2AK3|FOXP3|GATA6|GCK|GLIS3|GLUD1|HADH|HNF1A|HNF1B|HNF4A|IER3IP1|INS|INSR|KCNJ11|NEUROD1|NEUROG3|PAX4|PDX1|PTF1A|RFX6|SLC2A2|WFS1|ZFP57	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000508585.1	Genetic Services Laboratory University of Chicago Illinois 14D0917593 508585	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|GPR56|KIAA1279|LAMC3|LARGE|OCLN|PAFAH1B1|POMGNT1|POMT1|POMT2|RAB18|RAB3GAP1|RAB3GAP2|RELN|RTTN|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|WDR62	Deletion/duplication analysis	Microarray	Buccal swab|Cord blood|Fetal blood|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000508586.1	Genetic Services Laboratory University of Chicago Illinois	ACTB|ACTG1|ARX|DCX|FKRP|FKTN|GPR56|KIAA1279|LAMC3|LARGE|OCLN|PAFAH1B1|POMGNT1|POMT1|POMT2|RAB18|RAB3G	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Cell culture|Cord blood|Fetal blood|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	14D0917593 508586	AP1|RAB3GAP2|RELN|RTTN|TUBA1A|TUBA8|TUBB2B|TUBB3|VLDLR|WDR62					
GTR000509244.1	Genetics Laboratory Shodair Children's Hospital Montana 27D0652530 509244	Chromosome 22	FISH-metaphase	Metaphase FISH	Cord blood|Fetal blood|Peripheral (whole) blood	Decline to answer	Yes
GTR000500919.1	Heart Institute Diagnostic Lab Cincinnati Children's Hospital Medical Center Ohio 36D0656333 500919	TBX1	Mutation scanning of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	Decline to answer	No
GTR000053116.1	Human Genetics Laboratory, Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 53116	22q11.2	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization|Fluorescence in situ hybridization	Amniocytes|Amniotic fluid|Buccal swab|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Paraffin block|Peripheral (whole) blood|Product of conception (POC)	No	Yes
GTR000515046.3	Illumina Clinical Services Laboratory	NR	NR	NR	NR	NR	NR

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Illumina California 05D1092911 515046						
GTR000508888.1	Michigan Medical Genetics Laboratories University of Michigan Michigan 23D0366712 508888	Human genome	Deletion/duplication analysis	Microarray|Real Time Quantitative PCR	Peripheral (whole) blood	Decline to answer	Yes
GTR000500635.2	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500635	22q11.2	FISH-metaphase	Other	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Dried blood spot (DBS) card|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	No	Yes
GTR000335353.3	MitoMed Diagnostic Laboratory UCI California 05D1034314 335353	TBX1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	Buccal swab|Cell culture|Cord blood|Frozen tissue|Isolated DNA|Peripheral (whole) blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000501119.2	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 501119	ADA|CD3D|CD3E|DCLRE1C|FOXN1|IL2RG|IL7R|JAK3|LIG4|NHEJ1|ORAI1|PNP|PTPRC|RAG1|RAG2|RMRP|STAT5B|STIM1|TBX1|ZAP70	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	NR	Decline to answer	Yes
GTR000509589.1	Molecular Genetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 509589	TBX1	Sequence analysis of the entire coding region	Bi-directional Sanger Sequence Analysis	NR	Decline to answer	Yes
GTR000505776.3	PreventionGenetics PreventionGenetics Wisconsin 52D2065132 505776	ACVR2B|AHI1|ANKS6|ARL13B|ARL6|B9D1|B9D2|BBS1|BBS10|BBS12|BBS2|BBS4|BBS5|BBS7|BBS9|C5orf42|CC2D2A|CCDC103|CCDC11|CCDC114|CCDC39|CCDC40|CEP164|CEP290|CEP41|CFTR|DNAAF1|DNAAF2|DNAAF3|DNAH11|DNAH5|DNAI1|DNAI2|DNAL1|FOXH1|GDF1|GLIS2|HEATR2|INPP5E|INVS|IQCB1|KIF7|LEFTY2|LRRC6|MKKS|MKS1|NEK8|NKX2-5|NME8|NODAL|NPHP1|NPHP3|NPHP4|OFD1|RPGR|RPGRIP1L|RSPH4A|RSP	Deletion/duplication analysis|Sequence analysis of the entire coding region	Comparative Genomic Hybridization|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Amniocytes|Amniotic fluid|Bone marrow|Buccal swab|Cell culture|Cell-free DNA|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Product of conception (POC)|Saliva|Skin|Sputum|White blood	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H9|SDCCAG8|TCTN1|TCTN2|TCTN3|TMEM138|TMEM216|TMEM231|TMEM237|TMEM67|TRIM32|TTC21B|TTC8|WDPCP|WDR19|ZIC3|ZNF423			cell prep		
GTR000504359.1	Quest Diagnostics Nichols Institute Chantilly NR Virginia 49D0221801 504359	22q11.21	FISH-metaphase	Other	Peripheral (whole) blood	No	Yes
GTR000506041.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506041	22q11.21	FISH-metaphase	Other	Amniocytes|Chorionic villi|Peripheral (whole) blood	No	Yes
GTR000501985.3	Sequenom Center for Molecular Medicine – San Diego Sequenom CMM California 05D2015356 501985	1p36|22q11.2|5p15.2|Chromosome 13|Chromosome 15|Chromosome 16|Chromosome 18|Chromosome 21|Chromosome 22|Sex chromosome X|Sex chromosome Y	Targeted variant analysis	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes
GTR000509336.2	Transgenomic Transgenomic Connecticut	ABAT|ABCC8|ACOX1|ACY1|ADCK3|ADSL|AGA|AHI1|ALDH4A1|ALDH5A1|ALDH7A1|ALG1|ALG11|ALG12|A	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	07D0995237 509336	LG13|ALG2|ALG3|ALG6|ALG8|ALG9|AMT|ARFGEF2|ARG1|ARHGEF9|ARL13B|ARSA|ARSB|ARX|ASPA|ASPM|ATIC|ATP1A2|ATP2A2|ATP5A1|ATP6AP2|ATP6V0A2|ATP7A|ATPAF2|ATR|ATRX|B4GALT1|BCKDK|BCS1L|BRAF|BTD|C12orf57|C12orf65|CACNA1A|CACNA1H|CACNB4|CASK|CASR|CBL|CC2D2A|CCDC88C|CDK5RAP2|CDKL5|CDON|CENPJ|CEP152|CEP290|CHD2|CHRNA2|CHRNA4|CHRNB2|CLCN2|CLN3|CLN5|CLN6|CLN8|CNTN2|CNTNAP2|COG1|COG4|COG5|COG6|COG7|COG8|COL18A1|COL4A1|COQ2|COQ9|COX10|COX15|CPT2|CSTB|CTSA|CTSD|CUL4B|DCX|DDC|DDOST|DEPDC5|DHCR7|DLD|DOLK|DPAGT1|DPM1|DPM3|DPYD|DYRK1A|EFHC1|EIF2B1|EIF2B2|EIF2B3|EIF2B4|EIF2B5|EMX2|EOMES|EPM2A|ETFA|ETFB|ETFDH|FGD1|FGF8|FGFR3|FH|FKRP|FKTN|FLNA|FOLR1|FOXG1|FUCA1|GABRA1|GABRB3|GABRD|GABRG2|GALC|GALNS|GAMT|GATM|GCDH|GCSH|GFAP|GLB1|GLDC|GLI2|GLI3|GLRA1|GLRB|GLUD1|GLUL|GNE|GNPTAB|GNPTG|GNS|GOSR2|GPC3|GPHN|GPR56|GPR98|GRIA3|GRIN1|GRIN2A|GRIN2B|G					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		USB|HERC2|HEXA|HEXB|HGSNAT|HPD|HRAS|HSD17B10|HSD17B4|HYAL1|IDS|IDUA|INPP5E|IQSEC2|KAT6B|KCNA1|KCNJ10|KCNJ11|KCNMA1|KCNQ2|KCNQ3|KCNT1|KCTD7|KDM5C|KIAA1279|KMT2D|KRAS|L2HGDH|LAMA2|LARGE|LBR|LGI1|LRPPRC|MAGI2|MAGT1|MAP2K1|MAP2K2|MAPK10|MBD5|MCOLN1|MCPH1|ME2|MECP2|MED17|MEF2C|MFSD8|MGAT2|MGME1|MLC1|MMACHC|MOCS1|MOCS2|MOGS|MPDU1|MPI|MTHFR|MTR|MTRR|NAGLU|NDE1|NDUFA1|NDUFA2|NDUFAF6|NDUFS1|NDUFS3|NDUFS4|NDUFS7|NDUFS8|NDUFV1|NEU1|NF1|NHLRC1|NIPBL|NOTCH3|NPC1|NPC2|NPHP1|NRAS|NRXN1|OFD1|OPHN1|PAFAH1B1|PAK3|PANK2|PAX6|PC|PCDH19|PCNT|PDHA1|PDHX|PDSS1|PDSS2|PEX1|PEX10|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGK1|PGM1|PHF6|PHGDH|PIGV|PLA2G6|PLCB1|PLP1|PMM2|PNKP|PNPO|POLG|POMGNT1|POMT1|POMT2|PPT1|PQBP1|PRICKLE1|PRICKLE2|PRODH|PRRT2|PSAP|PSAT1|PTCH1|PTPN11|QDPR|RAB39B|RAB3GAP1|RAF1|RAI1|RARS2|RBFOX					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1|RELN|RFT1|RNASEH2A|RNASEH2B|RNASEH2C|RPGRIP1L|SAMHD1|SCARB2|SCN1A|SCN1B|SCN2A|SCN4A|SCN8A|SCN9A|SCO2|SDHA|SERPINI1|SETBP1|SGSH|SHH|SHOC2|SIX3|SLC16A2|SLC17A5|SLC19A3|SLC1A3|SLC25A15|SLC25A19|SLC25A22|SLC2A1|SLC35A1|SLC35A2|SLC35C1|SLC46A1|SLC4A10|SLC6A5|SLC6A8|SLC9A6|SMC1A|SMC3|SMPD1|SMS|SNAP29|SOS1|SPRED1|SPTAN1|SRD5A3|SRPX2|STIL|STXBP1|SUCLA2|SUMF1|SUOX|SURF1|SYN1|SYNGAP1|SYP|TACO1|TBC1D24|TBX1|TCF4|TGIF1|TMEM165|TMEM216|TMEM67|TMEM70|TPP1|TREX1|TSC1|TSC2|TSEN2|TSEN34|TSEN54|TUBA1A|TUBA8|TUBB2B|TUSC3|UBE3A|VDAC1|VPS13A|VPS13B|VRK1|WDR62|ZEB2|ZIC2					
GTR000305767.1	UW Cytogenetic Services University of Wisconsin - Madison / WSLH Wisconsin 52D0669558 305767	22q11.2	FISH-interphase|FISH-metaphase|Deletion/duplication analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray	Amniocytes|Amniotic fluid|Cord blood|Fibroblasts|Peripheral (whole) blood	Decline to answer	Yes

NR = Not reported

Table D-11. Genetic tests for Williams syndrome

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000019597.1	Center for Human Genetics Laboratory University Hospitals - University Hospitals Laboratory Service Foundation Ohio 36D0656024 19597	WBSCR	FISH-metaphase	Other	Amniocytes|Amniotic fluid|Chorionic villi|Cord blood|Cystic hygroma fluid|Fetal blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000206883.1	Center for Human Genetics, Inc Massachusetts 22D0650242 206883	7q11.23	FISH-metaphase	Fluorescence in situ Hybridization (FISH)	Not reported	Decline to answer	No
GTR000295229.2	Cincinnati Children's Cytogenetics Laboratory Cincinnati Children's Hospital Medical Center Ohio 36D0656333 295229	7q11.23	FISH-metaphase	Microduplication Metaphase FISH analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral (whole) blood|Product of conception (POC)	Decline to answer	Yes
GTR000295231.2	Cincinnati Children's Cytogenetics Laboratory Cincinnati	7q11.23	FISH-metaphase	Microdeletion Metaphase FISH analysis	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Peripheral	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Children's Hospital Medical Center Ohio 36D0656333 295231				(whole) blood|Product of conception (POC)		
GTR000500936.1	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 500936	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|C21orf33|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGD	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		H|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|G6PD|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|HTT|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000503671.2	Courtagen Diagnostics Laboratory Courtagen Life Sciences Massachusetts 22D2035168 503671	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ACACA|ACAD8|ACAD9|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACO2|ACSF3|ACSL4|ADSL|AFG3L2|AGK|AGPS|AGXT|AIFM1|AK2|AKAP10|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANKRD26|APTX|ARMS2|ASS1|ATIC|ATP5E|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BCKDHA|BCKDHB|BCKDK|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDKL5|CFTR|CHAT|CHRNA4|CHRNB2|CISD2|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|COA5|COMT|COQ2|COQ6|COQ9|COX14|COX15|COX4I2|COX6B1|COX7B|CPOX|CPS1|CPT1A|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDC|DGUOK|DHODH|DHTKD1|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNM1L|DTNBP1|EARS2|ELAC2|ELN|ENO3|ETFA|ETFB|ETFDH|ETHE1|FARS2|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FXN|G6PC|GAD1|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GF	Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)	Cord blood|Fresh tissue|Frozen tissue|Isolated DNA|Peripheral (whole) blood|Plasma|Product of conception (POC)|Saliva|Serum|Skin|Urine|White blood cell prep	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		M1|GK|GLDC|GLRA1|GLRX5|GLUD1|GLYCTK|GNAS|GNPAT|GPD1|GPD2|GPI|GPX1|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HAGH|HARS|HARS2|HCCS|HIBCH|HK1|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B2|HSPD1|HTRA2|IDH1|IDH2|IDH3B|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|L2HGDH|LDHA|LDHB|LIAS|LRPPRC|LRRK2|MAOA|MCCC1|MCCC2|MCEE|ME2|MECP2|MED23|MEN1|MFN2|MFSD8|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCS1|MOCS2|MOGS|MPC1|MPV17|MRPL3|MRPS16|MRPS22|MTFMT|MTHFD1|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NCOA4|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB3|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NFU1|NME1|NNT|NOS3|NRXN1|NT5C3A|NUBPL|OAT|OGDH|OGG1|OPA1|OPA3|OTC|OXCT1|PAH|PANK2|PARK2|PARK7|PC|PCBD1|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX11B|PEX12|PEX13|PEX14|PEX16|PEX1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		9|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHYH|PHYKPL|PINK1|PKLR|PNKD|PNMT|PNPT1|POLG|POLG2|PPARGC1B|PPOX|PPT1|PRODH|PTS|PUS1|PYCR1|QDPR|RARS2|REEP1|RMND1|RNASEL|RPL35A|RPS14|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SOD1|SOD2|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|SURF1|TAP1|TAT|TAZ|TCIRG1|TDP1|TIMM8A|TK2|TMEM126A|TMEM70|TP53|TPH2|TPP1|TRMU|TSFM|TTC19|TUFM|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|WFS1|WWOX|XPNPEP3|YARS2					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
GTR000325588.1	Cytogenetics and Molecular Genetics Laboratory Mercy St. Vincent Medical Center Ohio 36D0965797 325588	7q11.23	FISH-metaphase	FISH	Peripheral (whole) blood	Decline to answer	Yes
GTR000509084.1	Cytogenetics and Molecular Pathology Laboratory DLP Marquette General Hospital, LLC Michigan 23D1062707 509084	ELN	FISH-interphase|FISH-metaphase|Karyotyping	FISH|FISH|G-banding	Buccal swab|Cord blood|Fetal blood|Peripheral (whole) blood	No	Yes
GTR000013532.1	Cytogenetics Laboratory Indiana University School of Medicine Indiana 15D0647198 13532	7q11.23	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization (FISH)|Fluorescence in situ hybridization (FISH)	Amniocytes|Amniotic fluid|Bone marrow|Chorionic villi|Cord blood|Cystic hygroma fluid|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin|Urine	Decline to answer	Yes
GTR000032607.1	Cytogenetics Laboratory SUNY Upstate Medical University New York 33D0654590	7q11.23	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Cord blood|Fetal blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of	Yes	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	32607				conception (POC)|Skin		
GTR000500042.2	Cytogenetics Laboratory ARUP Laboratories, Inc. Utah 46D0523979 500042	15q11.2-q13|17p11.2|17p13.3|22q11.2|22q13.3|4p16.3|5p15.2|7q11.23|Xp22.3|Yp11.3	FISH-metaphase	FISH-metaphase	Amniocytes|Amniotic fluid|Cell culture|Chorionic villi|Cord blood|Fetal blood|Fibroblasts|Peripheral (whole) blood|Product of conception (POC)|Skin	Decline to answer	Yes
GTR000501846.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501846	7q11.23	FISH-metaphase	Fluorescence In Situ Hybridization	Cord blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood	No	No
GTR000501847.1	Emory Genetics Laboratory Emory University School of Medicine Georgia 11D0683478 501847	7q11.23	FISH-metaphase	Fluorescence In Situ Hybridization	Cell culture|Fibroblasts	No	No
GTR000509372.9	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509372	ABCC6|ACTA2|ACVR1|ADAMTS2|ATP6V0A2|CBS|CHST14|COL11A1|COL1A1|COL1A2|COL2A1|COL3A1|COL4A1|COL5A1|COL5A2|ELN|FBLN5|FBN1|FBN2|FKBP14|MYLK|NOTCH1|PKD2|PLOD1|PRDM5|SLC2A10|SLC39A13|SMAD3|TGFBR1|TGFBR2|ZNF469	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000509394.8	Fulgent Clinical	EFEMP2|ELN|FBLN5|FLC	Deletion/duplication	Next-Generation	Buccal swab|Isolated	Decline to	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Diagnostics Lab Fulgent Diagnostics California 05D2043189 509394	N|LTBP4|SERPINA1|TSC1|TSC2	analysis|Sequence analysis of the entire coding region	(NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	DNA|Peripheral (whole) blood|Saliva	answer	
GTR000509435.8	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 509435	ABCA3|ACVRL1|AP3B1|ASCL1|BDNF|BLOC1S3|BLOC1S6|BMPR2|CCDC39|CCDC40|CFTR|DNAAF1|DNAAF2|DNAH11|DNAH5|DNAI1|DNAI2|DNAL1|DTNBP1|EDN3|EFEMP2|ELN|ENG|FBLN5|FLCN|GDNF|HPS1|HPS3|HPS4|HPS5|HPS6|LTBP4|MUC5B|NME8|PHOX2B|RET|RSPH4A|RSPH9|SCNN1A|SCNN1B|SCNN1G|SERPINA1|SFTPA1|SFTPA2|SFTPB|SFTPC|SMAD9|TERC|TERT|TSC1|TSC2	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No
GTR000510915.7	Fulgent Clinical Diagnostics Lab Fulgent Diagnostics California 05D2043189 510915	AAAS|AARS2|AASS|ABAT|ABCB6|ABCB7|ABCC8|ABCC9|ABCD1|ABCD3|ACACA|ACACB|ACAD8|ACAD9|ACADL|ACADM|ACADS|ACADSB|ACADVL|ACAT1|ACAT2|ACHE|ACLY|ACO2|ACSF3|ACSL4|ACSL5|ACSM3|ADSL|AFG3L2|AGK|AGPS|AGXT|AGXT2|AIFM1|AK2|AKAP10|AKR7A2|AKT1|AKT2|ALAS2|ALDH18A1|ALDH2|ALDH3A2|ALDH4A1|ALDH5A1|ALDH6A1|ALDH7A1|AMACR|AMT|ANK2|ANKRD26|APTX|ARMS2|AS3MT|ASS1|ATIC|ATP10D|ATP5E|ATP5SL|ATP7B|ATP8B1|ATPAF2|ATXN7|AUH|BAX|BCAT1|BCAT2|BCKD	Deletion/duplication analysis|Sequence analysis of the entire coding region	Next-Generation (NGS)/Massively parallel sequencing (MPS)|Next-Generation (NGS)/Massively parallel sequencing (MPS)	Buccal swab|Isolated DNA|Peripheral (whole) blood|Saliva	Decline to answer	No

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		HA|BCKDHB|BCL2|BCS1L|BOLA3|C10orf2|C12orf65|CACNA1A|CACNA1S|CACNA2D1|CASP8|CDC42BPB|CDKL5|CFTR|CHAT|CHDH|CHRNA4|CHRNB2|CISD2|CKM|CLCN1|CLCN2|CLCN5|CLCN7|CLCNKB|CLN3|CLN5|CLN6|CLN8|CLYBL|CNR1|COA5|COMT|COQ2|COQ4|COQ5|COQ6|COQ9|COX10|COX15|COX4I1|COX4I2|COX6B1|COX7A2|CPOX|CPS1|CPT1A|CPT1B|CPT2|CTSD|CYB5A|CYB5R3|CYBA|CYBB|CYCS|CYP11A1|CYP11B1|CYP11B2|CYP24A1|CYP27A1|CYP27B1|D2HGDH|DARS2|DBT|DDAH1|DDC|DECR1|DGUOK|DHODH|DIABLO|DISC1|DLAT|DLD|DMGDH|DMPK|DNAJC19|DNAJC5|DNM1L|DTNBP1|EARS2|ECI1|ECSIT|ELAC2|ELN|ENO1|ENO3|ETFA|ETFB|ETFDH|ETHE1|FAAH|FARS2|FASN|FASTKD2|FBP1|FECH|FH|FOLR1|FOXC1|FOXG1|FOXRED1|FPGS|FTH1|FXN|G6PC|G6PD|GAD1|GAD2|GALC|GARS|GATM|GCDH|GCK|GCSH|GDAP1|GFER|GFM1|GK|GLDC|GLO1|GLRA1|GLRX5|GLS|GLUD1|GLYCTK|GNAS|GNPAT|GPAM|GPD1|GPD2|GPI|GPX1|GPX4|GYS1|GYS2|H6PD|HADH|HADHA|HADHB|HARS|HARS2|HCCS|HIBCH|HIGD2A|HK1|HK2|HLCS|HMGCL|HMGCS2|HOGA1|HSD17B10|HSD17B4|HSD3B1|HSD3					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		B2|HSPA9|HSPB7|HSPD1|HTRA2|HTT|IDE|IDH1|IDH2|IDH3B|IMMP2L|IMMT|INSR|ISCU|IVD|KARS|KCNA1|KCNE1|KCNE2|KCNH2|KCNJ11|KCNJ2|KCNQ1|KCNQ2|KCNQ3|KIF1B|KRT5|KYNU|L2HGDH|LARS2|LDHA|LDHB|LETM1|LIAS|LRPPRC|LRRK2|MAOA|MAOB|MARS2|MAVS|MCCC1|MCCC2|MCEE|MDH1|MECP2|MED23|MEN1|MFN2|MFSD8|MGLL|MGST3|MLYCD|MMAA|MMAB|MMACHC|MMADHC|MOCOS|MOCS1|MOCS2|MOGS|MRPL3|MRPL48|MRPS16|MRPS22|MRRF|MTCH2|MTFMT|MTHFD1|MTHFD1L|MTHFS|MTO1|MTPAP|MTRR|MUT|MUTYH|NAGS|NARS2|NDUFA1|NDUFA10|NDUFA11|NDUFA12|NDUFA13|NDUFA2|NDUFA4|NDUFA6|NDUFA7|NDUFA8|NDUFA9|NDUFAF1|NDUFAF2|NDUFAF3|NDUFAF4|NDUFB1|NDUFB3|NDUFB6|NDUFB9|NDUFC2|NDUFS1|NDUFS2|NDUFS3|NDUFS4|NDUFS5|NDUFS6|NDUFS7|NDUFS8|NDUFV1|NDUFV2|NDUFV3|NFU1|NIPSNAP1|NIPSNAP3A|NLRX1|NME1|NOS3|NPL|NRXN1|NTHL1|NUBPL|OAT|OGG1|OPA1|OPA3|OTC|OXCT1|PACRG|PAH|PAK7|PANK2|PARK2|PARL|PARP1|PC|PCCA|PCCB|PCK1|PCK2|PDHA1|PDHB|PDHX|PDP1|PDSS1|PDSS2|PDX1|PEX1|PEX10|PEX1					

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
		1B|PEX12|PEX13|PEX14|PEX16|PEX19|PEX2|PEX26|PEX3|PEX5|PEX6|PEX7|PGAM2|PGK1|PHB|PHYH|PKLR|PMPCA|PNKD|PNMT|POLG|POLG2|POLRMT|PPARGC1A|PPARGC1B|PPOX|PPT1|PREPL|PRODH|PTGES2|PTS|PUS1|PYCR1|QDPR|RAB11FIP5|RARS2|REEP1|RNASEL|RPL35A|RRM2B|RSPH9|RYR1|RYR2|SACS|SARDH|SARS2|SCN1A|SCN1B|SCN2A|SCN4A|SCN5A|SCO1|SCO2|SCP2|SDHA|SDHAF1|SDHAF2|SDHB|SDHC|SDHD|SECISBP2|SHMT1|SIRT1|SIRT3|SIRT5|SLC16A1|SLC19A2|SLC22A4|SLC22A5|SLC25A12|SLC25A13|SLC25A15|SLC25A19|SLC25A20|SLC25A22|SLC25A3|SLC25A38|SLC25A39|SLC25A4|SLC27A4|SLC2A1|SLC3A1|SLC6A8|SPAST|SPG20|SPG7|SPR|SPTLC2|STAR|SUCLA2|SUCLG1|SUGCT|SUOX|TACO1|TAP1|TAT|TAZ|TCIRG1|TDP1|TFAM|TFB1M|TIMM44|TIMM8A|TK2|TMEM126A|TMEM70|TOMM40|TOP1MT|TP53|TPH2|TPI1|TPP1|TRMU|TSFM|TSPO|TST|TTC19|TUFM|TXN2|TXNRD2|TYMP|UBE3A|UCP1|UCP2|UCP3|UNG|UQCRB|UQCRQ|UROS|USP24|WFS1|WWOX|XPNPEP3|YARS2					
GTR000053130.2	Human Genetics Laboratory,	7q11.23	FISH-interphase|FISH-metaphase	Fluorescence in situ hybridization|Fluorescence in	Fetal blood|Peripheral	No	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Munroe-Meyer Institute University of Nebraska Medical Center Nebraska 28D0454363 53130			situ hybridization	(whole) blood		
GTR000500654.2	Michigan State University Clinical Genetics Laboratory Michigan State University Michigan 23D0650879 500654	7q11.23	FISH-metaphase	Other	Amniocytes|Bone marrow|Cell culture|Chorionic villi|Fetal blood|Fibroblasts|Fresh tissue|Peripheral (whole) blood|Product of conception (POC)|Skin	No	Yes
GTR000504365.1	Quest Diagnostics Nichols Institute Chantilly Virginia 49D0221801 504365	7q11.23	FISH-metaphase	Other	Peripheral (whole) blood	No	Yes
GTR000506049.2	Quest Diagnostics Nichols Institute San Juan Capistrano California 05D0643352 506049	7q11.23	FISH-metaphase	Other	Amniocytes|Chorionic villi|Peripheral (whole) blood	No	Yes
GTR000305785.1	UW Cytogenetic Services University of Wisconsin -	7q11.23	FISH-interphase|FISH-metaphase|Deletion/duplication analysis	Fluorescence In Situ Hybridization|Fluorescence In Situ Hybridization|Microarray	Amniocytes|Amniotic fluid|Cord blood|Fibroblasts|Peripheral (whole) blood	Decline to answer	Yes

GTR Test ID	Lab Name of Institution State CLIA Number Lab Test ID	Target Chromosomal Regions or Genes	Method Categories	Methods	Specimen Source	Genetic Counseling Required Pre-Test or Post-Test	Proficiency Testing Performed
	Madison / WSLH Wisconsin 52D0669558 305785						

Appendix E. Ongoing Clinical Trials

Appendix E-1. Ongoing clinical trials

Identifier	Sponsor	Design	Purpose	Outcomes	Start Date and Expected Completion Date
Research and characterization of new genes involved in Intellectual Disability (GeneDefi) NCT01867554	Institute National de la Sante Et of Medical Research, France	Prospective observational cohort study enrolling 2,500 patients with intellectual disability or psycho-motor retardation, 5,000 parents or unaffected siblings, and 1,000 affected siblings. Researchers will collect urine samples for processing.	To identify genes involved in intellectual disability with SNP-arrays, next generation sequencing and other newer methods and establish genotype-phenotype correlations.	Outcomes include the number of participants with a genetic cause identified and genotype-phenotype correlations over a 5-year followup period.	Start date: December 2012 End date: December 2022
Inter-regional project of the great western exploration approach for exome molecular causes severe intellectual disability isolated or syndromic (HUGODIMS) NCT02136849	Nantes University Hospital, France	Prospective observational study of 225 patients with severe or moderate intellectual disability with isolated or syndromic presentation with negative fragile X and CGH results.	To evaluate high-throughput sequencing (NGS), to compare the exome of the child to the parent, and its contribution in the diagnostic management of patients with intellectual disability.	Outcomes to be assessed include the number of patients for whom de novo mutations are identified, the number of mutations compatible with the mode of recessive autosomal recessive or X-linked chromosome, and the number of de novo mutations (loss of function, missense, or indels) probably pathogens identified that are not known to be involved with the intellectual disability genes.	Start date: July 2014 Completion date: January 2017

Identifier	Sponsor	Design	Purpose	Outcomes	Start Date and Expected Completion Date
The psychiatric and cognitive phenotypes in velocardiofacial syndrome, Williams syndrome, and fragile X syndrome characterization, treatment, and examining the connection to developmental and molecular factors NCT00768820	Rabin Medical Center, Israel	Prospective interventional case series study of the efficacy and safety of methylphenidate, fluoxetine, and risperidone in 400 patients with a chromosomal deletion on FISH examination.	To investigate the psychiatric and cognitive phenotypes in velocardiofacial syndrome, Williams syndrome, and Fragile X syndrome, their response to treatment, and its relationship with molecular factors.	Outcomes to be assessed include psychological measures, molecular tests, and responsiveness to medical treatment at two years followup.	Start date: May 2001 Completion date: December 2012 Status is unknown; last NCT update was May 2012
Acquisition of saliva samples for validation as a specimen type in the Esoteric Genetic Laboratories fragile X assay NCT01616589	Esoterix Genetic Laboratories, LLC, Burlington, NC, USA	Prospective cohort study of 100 patients previously identified as have fragile X intermediate, permutation (carrier) or full mutation (affected).	To determine whether saliva samples can be used as an alternative sample type to test for fragile X syndrome.	Outcome is fragile X diagnosis based on saliva sample.	Start date: not reported. Completion date: December 2012 Status unknown; last NCT update was June 2012
SynapDx autism spectrum disorder gene expression analysis study. NCT01810341	SynapDx Corp., Lexington, MA, USA	Prospective observational cohort study of 880 patients between 18 and 60 months of age referred to a developmental evaluation center for evaluation of a possible developmental disorder.	To develop an algorithm to classify blood RNA gene expression patterns to maximize agreement between the classification and a clinical assessment of the presence or absence of autism spectrum disorder	Outcomes to be assessed include RNA expression in peripheral blood	Start date: March 2013 Completion date: September 2014 Study has been completed
Study of correlation between expression of proteins that are essential for embryonic brain development and neurodevelopmental outcomes at 2 years of age in premature infants. NCT01686685	Hadassah Medical Organization, Israel	Prospective observational cohort study of 400 premature infants	Because premature birth is associated with an increased risk of autism, this study plans to measure changes in the expression of 3 different proteins that may be affected by changes in oxygen level at birth. Investigators will study the interaction between changes in protein levels in the first few days after premature birth and a diagnosis of ASD at 2 years of age.	Outcomes to be assessed include diagnoses of ASD at 2 years, and corrected age and language or cognitive delay at 2 years corrected age	Start date: April 2012 Completion date: not reported Status is unknown; last NCT update was September 2012

Identifier	Sponsor	Design	Purpose	Outcomes	Start Date and Expected Completion Date
Exosomal MicroRNA expression in children with autism spectrum disorder. NCT01749670	State University of New York – Upstate Medical Center, New York State, USA	Case-control study of 40 children. Cases include children 4–17 years of age with ASD versus age- and gender-matched controls with neuropsychological developmental patterns.	To identify small RNA particles that regulate gene expression in autism spectrum disorder and determine which miRNA patterns may improve our understanding and diagnosis of ASD.	The outcome to be assessed is the correlation between miRNA expression and medical/ neuropsychological characteristics of study participants.	Start date: January 2013 Completion date: January 2014 Study has been completed
Early detection of autism spectrum disorder in children NCT01646866	St. Louis University, St. Louis, Missouri, USA	Prospective observational cohort study of 50 biological siblings of a child with a diagnosis of ASD. The siblings will display red flags for autism through 1 year of age.	To identify infants at risk for autism less younger than 1 year of age to improve outcomes through early diagnosis and treatment. Correlation between clinical screening and genetic screening will be evaluated.	Outcomes to be assessed include the percentage of siblings with a high-risk genetic score on the ARISK genetic test who have a diagnosis of ASD at 24 months and the percentage of siblings who failed the Red Flags for Communication Scale at 12 months who have a diagnosis of ASD at 24 months	Start date: December 2010 Completion date: December 2016
Autism and glutamatergic synapse: Research of genetic mutations and identification of clinical and neurophysiological markers. NCT01770548	University Hospital, Tours, France	Open-label, nonrandomized, parallel-assignment study enrolling 600 patients with a pervasive developmental disorder. Patients in the experimental arm will undergo an analysis of the glutamate synapse in autism, DNA will be collected, and auditory evoked potentials will be measured.	To analyze 50 genes in the glutamate synapse.	Outcomes to be assessed include an analysis of 50 genes encoding proteins of the NMDA receptor complex, chosen according to their chromosomal localization located in hotspots depicted by genome-wide screens in a large population of autistic children.	Start date: January 2009 Completion date: January 2011 Status unknown; last NCT update was January 2013

Identifier	Sponsor	Design	Purpose	Outcomes	Start Date and Expected Completion Date
Use of pharmacogenetics in the treatment of children with ASD NCT00859664	Assaf-Harofeh Medical Center, Israel	Prospective observational cohort study of 500 patients older than 3 years of age diagnosed with autism and treated with neuroleptics.	To map the different types of cytochrome P450 gene in children with autism to identify those for whom psychiatric drug treatment is successful and to prevent adverse drug reactions.	To evaluate the relationship between genotypes of the cytochrome P450 (CYP) 2D6 gene and response to treatment with psychiatric medications.	Start date: March 2009 Completion date: July 2011 Status unknown; last NCT update was March 2009

ASD=autism spectrum disorder; CGH=comparative genomic hybridization; FISH=fluorescent in situ hybridization; NCT=National Clinical Trials database; NGS=next-generation sequencing; SNP=single nucleotide polymorphism

Appendix F. Excluded Studies Based on Review of Full-Length Articles

Not a disorder of interest

Barcia G, Chemaly N, Gobin S, et al. Early epileptic encephalopathies associated with STXBP1 mutations: Could we better delineate the phenotype? Eur J Med Genet. 2014 Jan;57(1):15-20.

Cody JD, Hasi M, Soileau B, et al. Establishing a reference group for distal 18q-: Clinical description and molecular basis. Hum Genet. 2014 Feb;133(2):199-209. PMID: 24092497.

Del Giudice E, Macca M, Imperati F, et al. CNS involvement in OFD1 syndrome: a clinical, molecular, and neuroimaging study. Orphanet J Rare Dis. 2014 May 10;9(1):74.

Not a study design of interest

Carayol J, Tores F, Konig IR, et al. Evaluating diagnostic accuracy of genetic profiles in affected offspring families. Stat Med. 2010 Sep 30;29(22):2359-68. PMID: 20623818.

Not a genetic test of interest

Mahajan S, Kaur A, Singh JR. Cytogenetic investigations in mentally challenged individuals. Int J Hum Genet. 2011 Jun;11(2):93-8.

Not a US-based economic study

Regier DA, Friedman JM, Marra CA. Value for money? Array genomic hybridization for diagnostic testing for genetic causes of intellectual disability. Am J Hum Genet. 2010 May 14;86(5):765-72. PMID: 20398885.

Antonanzas F, RodriguezIbeas R, Hutter MF, et al. Genetic testing in the European Union: does economic evaluation matter? Eur J Health Econ. 2012 Oct;13(5):651-61. http://dx.doi.org/10.1007/s10198-011-0319-x. PMID: 21598012.

A narrative review

Freitag CM, Staal W, Klauck SM, et al. Genetics of autistic disorders: review and clinical implications. Eur Child Adolesc Psychiatry. 2010 Mar;19(3):169-78. PMID: 19941018.

Appendix G. Abbreviations and Acronyms

AAIDD	American Association on Intellectual and Developmental Disabilities
aCGH	microarray-based comparative genomic hybridization
AHRQ	Agency for Healthcare Research and Quality
ASD	autism spectrum disorder
AUC	area under the curve
CDC	Centers for Disease Control and Prevention
CGH	comparative genomic hybridization
CLIA	Clinical Laboratory Improvement Amendments of 1988
CMA	chromosomal microarray analysis
CNV	copy number variation
DD	developmental disability
EPC	Evidence-based Practice Center
FDA	U.S. Food and Drug Administration
FISH	fluorescence in situ hybridization
GDD	global developmental delay
GTR	Genetic Testing Registry
ID	intellectual disability
IQ	intelligence
KI	Key Informant
LDT	laboratory-developed test
MLPA	multiplex ligation-dependent probe amplification
NCBI	National Center for Biotechnology Information
NGS	next-generation sequencing
OMIM	Online Mendelian Inheritance in Man
PCR	polymerase chain reaction
PT	proficiency testing
RCT	randomized controlled trial
ROC	receiver operator characteristic
SNP	single nucleotide polymorphism
WES	whole exome sequencing
WGS	whole genome sequencing

Appendix H. Appendix Reference List

1. Cambell NA, Reece JB, Mitchell LG. Biology. 5th ed. Menlo Park (CA): Benjamin Cummings; 1999.
2. Hartl DL, Jones EW. Genetics: analysis of genes and genomes. 5th ed. Sudbury (MA): Jones and Bartlett Publishers; 2001.
3. Genetics home reference: Down syndrome. Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/condition/down-syndrome. Accessed 2014 Sep 30.
4. Genetics home reference: Rett syndrome. Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/condition/rett-syndrome. Accessed 2014 Sep 30.
5. Genetics home reference: MECP2. Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/gene/MECP2. Accessed 2014 Sep 30.
6. Genetics home reference: glossary: copy number variation. Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/glossary=copynumbervariation. Accessed 2014 Sep 30.
7. Bickmore WA. Karyotype analysis and chromosome banding. In: Encyclopedia of life sciences. London (UK): Nature Publishing Group; 2001. p. 1-7. Also available: http://www.unioviedo.es/esr/pp/bandmethods.pdf.
8. Fluorescence In Situ Hybridization (FISH). National Human Genome Research Institute; 2011 Oct 13. http://www.genome.gov/10000206. Accessed 2014 Oct 02.
9. Beaudet AL. The utility of chromosomal microarray analysis in developmental and behavioral pediatrics. Child Dev. 2013 Jan-Feb;84(1):121-32. PMID: 23311723.
10. Theisen A. Microarray-based comparative genomic hybridization (aCGH). Nat Educ. 2008;1(1):45.
11. Microarray-based comparative genomic hybridization (array CGH). Rare Chromosome Disorder Support Group; 2013. 12 p. Also available: http://www.rarechromo.org/information/other/array%20cgh%20ftnw.pdf.
12. Genetics home reference: what are single nucleotide polymorphisms (SNPs)? Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/handbook/genomicresearch/snp. Accessed 2014 Sep 30.
13. LaFramboise T. Single nucleotide polymorphism arrays: a decade of biological, computational and technological advances. Nucleic Acids Res. 2009 Jul;37:4181-93. PMID: 19570852.
14. Sherry ST, Ward MH, Kholodov M, et al. dbSNP: the NCBI database of genetic variation. Nucleic Acids Res. 2001 Jan 1;29:308-11. PMID: 11125122.
15. Marian AJ. Sequencing your genome: what does it mean? Methodist Debakey Cardiovasc J. 2014 Jan-Mar;10:3-6. PMID: 24932355.
16. Genetics home reference: what advances are being made in DNA sequencing. Bethesda (MD): U.S. National Library of Medicine (NLM); 2014 Sep 29. http://ghr.nlm.nih.gov/handbook/genomicresearch/sequencing. Accessed 2014 Oct 01.
17. Medical genetics test details: whole exome sequencing. Houston (TX): Baylor College of Medicine. https://www.bcm.edu/research/medical-genetics-labs/test_detail.cfm?testcode=1500. Accessed 2014 Oct 01.
18. Rabbani B, Tekin M, Mahdieh N. The promise of whole-exome sequencing in medical genetics. J Hum Genet. 2014 Jan;59(1):5-15. PMID: 24196381.
19. Ng SB, Turner EH, Robertson PD, et al. Targeted capture and massively parallel sequencing of 12 human exomes. Nature. 2009 Sep 10;461(7261):272-6. PMID: 19684571.
20. Teer JK, Mullikin JC. Exome sequencing: the sweet spot before whole genomes. Hum Mol Genet. 2010 Oct 15;19:R145-51. PMID: 20705737.
21. Chorev M, Carmel L. The function of introns. Front Genet. 2012;3:55. PMID: 22518112.
22. Beachy, et al. Assessing genomic sequencing information for health care decision making: workshop summary. Washington (DC): National Academies Press; 2014.

23. Policy statement: points to consider in the clinical application of genomic sequencing. Bethesda (MD): American College of Medical Genetics and Genomics (ACMG); 2012 May 15. 4 p. Also available: https://www.acmg.net/StaticContent/PPG/Clinical_Application_of_Genomic_Sequencing.pdf.

24. Dewey FE, Grove ME, Pan C, et al. Clinical interpretation and implications of whole-genome sequencing. JAMA. 2014 Mar 12;311:1035-45. PMID: 24618965.

25. PCR fact sheet. Bethesda (MD): National Human Genome Research Institute (NHGRI); 2014 Jul 17. http://www.genome.gov/10000207. Accessed 2014 Sep 30.

26. Giasuddin AS. Polymerase chain reaction technique: fundamental aspects and applications in clinical diagnostics. J Islam Acad Sci. 1995;8(1):29-32.

27. Henegariu O, Heerema NA, Dlouhy SR, et al. Multiplex PCR: critical parameters and step-by-step protocol. Biotechniques. 1997 Sep;23(3):504-11. PMID: 9298224.

28. Real-Time qRT-PCR - Real-Time Quantitative Reverse Transcription PCR. Bethesda (MD): National Center for Biotechnology Information (NCBI). http://www.ncbi.nlm.nih.gov/projects/genome/probe/doc/TechQPCR.shtml. Accessed 2014 Oct 01.

29. Heid CA, Stevens J, Livak KJ, et al. Real time quantitative PCR. Genome Res. 1996 Oct;6(10):986-94. PMID: 8908518.

30. Stuppia L, Antonucci I, Palka G, et al. Use of the MLPA Assay in the Molecular Diagnosis of Gene Copy Number Alterations in Human Genetic Diseases. Int J Mol Sci. 2012;13(3):3245-76. PMID: 22489151.

31. MRC-Holland: multiplex ligation-dependent probe amplification (MLPA): copy number quantification and methylation detection [slide set]. Amsterdam (the Netherlands): MRC-Holland; 50 p. Also available: http://www.mlpa.com/WebForms/WebFormMain.aspx?Tag=_wl2zCji-rCGANQgZPuTixsEyIW1MscfzuKj2NDFYc-g.

32. Phillips T. The role of methylation in gene expression. Nat Educ. 2008;1(1):116.

33. Shen L, Waterland RA. Methods of DNA methylation analysis. Curr Opin Clin Nutr Metab Care. 2007 Sep;10:576-81. PMID: 17693740.

34. Sun F, Bruening W, Erinoff E, et al. Addressing challenges in genetic test evaluation. Evaluation frameworks and assessment of analytic validity Methods research report. (Prepared by the ECRI Institute Evidence-based Practice Center under Contract No. HHSA 290-2007-10063-I.) AHRQ publication no. 11-EHC048-EF. Rockville (MD): Agency for Healthcare Research and Quality (AHRQ); 2011 Jun. 107 p. Also available: http://www.ncbi.nlm.nih.gov/books/NBK56750/pdf/TOC.pdf.