Effective Health Care

Bone Marrow Transplant to Treat DOCK8 Deficiency
Nomination Summary Document

Results of Topic Selection Process & Next Steps

- The topic, *Bone Marrow Transplant to Treat DOCK8 Deficiency*, is not feasible for a full systematic review due to the limited data available for a review at this time.
- *Bone Marrow Transplant to Treat DOCK8 Deficiency* could potentially be considered for new research in comparative effectiveness.

Topic Description

Nominator(s): Individual

Nomination Summary: The nominator is a physician treating a patient with DOCK8 deficiency, systemic lupus erythematosus, and antiphospholipid syndrome. The nominator believes that more research is necessary to evaluate the effectiveness of bone marrow transplants, and more generally hematopoietic cell transplantations, to treat DOCK8 deficiency. The nominator also notes that this research is particularly necessary to understand the optimal treatment of patients with DOCK8 deficiency who have additional comorbidities. The nominator hopes that additional research or synthesis of the evidence related to DOCK8 deficiency will identify new treatment options and more broadly improve physician understanding of the optimal treatment of patients with multiple diseases.

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Population(s): Patients with DOCK8 deficiency
Intervention(s): Hematopoietic cell transplantation
Comparator(s): No transplant, supporting therapy
Outcome(s): Morbidity, mortality, hospitalizations, occurrence of adverse events, quality of life

Key Questions from Nominator: What is the safety and effectiveness of hematopoietic cell transplantation to treat DOCK8 deficiency as compared to other treatment options?

Considerations

- The topic meets Effective Health Care (EHC) Program appropriateness and some of the importance criteria. (For more information, see http://effectivehealthcare.ahrq.gov/index.cfm/submit-a-suggestion-for-research/how-are-research-topics-chosen/.)
- DOCK8 deficiency is a rare immune system disorder that was first identified in 2009 (as of 2010, only 32 cases had been identified worldwide). Previously, patients with the condition were typically

categorized as having hyper IgE syndrome, which is another immune system disorder with similar symptoms.

- DOCK8 deficiency is associated with comorbidities, including dermatologic conditions, recurrent pneumonia, recurrent viral infections, severe allergies, asthma, and malignancies such as squamous cell carcinomas.
- The treatment of DOCK8 deficiency may include hematopoietic stem cell transplantation in addition to the management of symptoms. One example is the use anti-inflammatory medications to treat the associated eczema.
- A literature scan for published trials related to the treatment of DOCK8 deficiency, identified individual case studies or non-controlled studies with small sample sizes.
- A search of clinicaltrails.gov identified two ongoing clinical trials:
    - NCT01176006 – Pilot study of reduced-intensity hematopoietic stem cell transplant of DOCK8 deficiency. Estimated completion date: None listed.
    - NCT01212055 – Apheresis of patients with immunodeficiency. Estimated completion date: None listed.
- There appears to be a limited amount of evidence comparing the effectiveness of different strategies; therefore as systematic review is not feasible at this time. Additional research could help to further support the development of an evidence base around the treatment of DOCK8 deficiency.