Effective Health Care
Enzyme Replacement Therapy for Lysosomal Storage Disease Nomination Summary Document

Results of Topic Selection Process & Next Steps

- Enzyme replacement therapy for lysosomal storage disease is not feasible for a full systematic review due to the limited data available at this time; however, it will be considered for a potential technical brief by the Effective Health Care Program.

- To see a description of a technical brief, please go to http://effectivehealthcare.ahrq.gov/index.cfm/research-for-policymakers-researchers-and-others/.

- If this topic is developed into a technical brief, key questions will be drafted and posted on the AHRQ Web site. To sign up for notification when this and other Effective Health Care Program topics are posted, please go to http://effectivehealthcare.ahrq.gov/index.cfm/join-the-email-list1/.

Topic Description

Nominator: Public payer

Nomination Summary: The nominator questions the effectiveness of enzyme replacement therapy (ERT) for the treatment of lysosomal storage diseases. The nominator is also interested in the long-term clinical outcomes associated with ERT.

Key Questions from Nominator: None

Considerations

- The topic meets EHC Program appropriateness and importance criteria. (For more information, see http://effectivehealthcare.ahrq.gov/index.cfm/submit-a-suggestion-for-research/how-are-research-topics-chosen/.)

- There are approximately 41 LSDs, and only a few are treatable with ERT. Typically, the infantile forms of these diseases are much more severe than the adult forms, often leading to mortality before the age of 5 years. Historical treatments for LSDs were primarily palliative before the advent of ERT.

- Enzyme replacement therapy is a relatively new treatment modality with approximately 10-15 years of usage primarily in Gaucher's and Fabry's diseases. A small amount of long-term data on effectiveness and harms of this treatment in these diseases has been published.
- For the other Lysosomal Storage Diseases, much of the literature is limited to small trials, case series, or case reports. Long-term data is not yet available for many of these diseases.

- No comprehensive evidence review was identified that addresses all of the current research in this area. Given the adequacy and volume of the current literature, this topic will be considered for a technical brief.