TEXAS Health and Human Services
Texas Department of State Health Services
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Commissioner

Newborn Screening ACT Sheet

Mucopolysaccharidosis Type 1 (MPS I)

Condition Description: MPS I is an autosomal recessive lysosomal storage disorder caused by pathogenic variants in the *IDUA* gene leading to deficient alpha-L-iduronidase activity. Symptoms can include umbilical and/or inguinal hernia, macrocephaly, macroglossia, hepatosplenomegaly, coarse facial features, cardiac abnormalities, respiratory abnormalities, and stiff joints. This deficiency leads to the accumulation of glycosaminoglycans (also known as mucopolysaccharides) in the lysosome resulting in cellular dysfunction. There is wide variability in severity and age of onset.

Please Take the Following Immediate Actions:

- ☐ **Consult with pediatric metabolic specialist.** (See attached list).
- ☐ **Contact family to inform them of the newborn screening result.** Newborns are expected to be **asymptomatic.** We provide a resource for this conversation: **Next steps after a positive Newborn Screening for MPS I**
- ☐ **Obtain initial labs:** urine glycosaminoglycans (GAGs) quantitative analysis (does not need to be a sterile sample, clean catch is fine) and serum alpha- L-iduronidase enzyme assay in leukocytes.
- ☐ **Collect repeat screen** (between 7-14 days of life) if the second screen has not been done.
- ☐ **FAX** lab results to **{SGUserFax}**

Diagnostic Evaluation: All infants with an abnormal MPS I newborn screening result need a clinical examination and lab evaluation with urine GAG analysis, enzyme testing, and *IDUA* genetic analysis. The diagnosis of MPS I is based on elevated urine GAGs and deficient alpha-L- iduronidase enzyme, along with genetic analysis.

Clinical Considerations: The prognosis is best for newborns who are diagnosed and treated quickly, prior to two years of age. There is no cure for this condition, but treatments can help to delay some symptoms and manage others. Without treatment, babies with severe MPS I will experience a progressive decline in cognitive function, worsening of symptoms, and a shorter lifespan.

Additional Information:

How to Communicate Newborn Screening Results
https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritabledisorders/resources/achdnc-communication-guide-newborn.pdf
MedlinePlus https://ghr.nlm.nih.gov/condition/mucopolysaccharidosis-type-i
National Organization for Rare Diseases www.rarediseases.org/rare-diseases/mucopolysaccharidoses
American College of Medical Genetics and Genomics Knowledge Nuggets
https://nccrcg.org/knowledge-nugget-series/