MAYO CLINIC LABORATORIES

Newborn Screening Act Sheet

Mucopolysaccharidoses Type I: Decreased Alpha-L-Iduronidase

Differential Diagnosis: Hurler syndrome (MPS IH), Hurler-Scheie syndrome (MPS IH/S), and Scheie syndrome (MPS IS)

Condition Description: Mucopolysaccharidoses type I (MPS I) is a lysosomal storage disorder (LSD) caused by a defect in alpha-L-iduronidase activity, resulting in accumulation of glycosaminoglycans (also known as mucopolysaccharides) within the lysosome. This accumulation results in cell enlargement and subsequent dysfunction. There is variability in severity and age of onset. MPS I is an autosomal recessive disorder.

You should take the following actions:

- Contact family to inform them of the newborn screening result and ascertain clinical status (umbilical and/or inguinal hernia, macrocephaly, macroglossia, hepatosplenomegaly, coarse facial features).
- Consult with genetic or metabolic specialist.
- Evaluate the newborn (presence of hernia, liver/spleen size, head size, cardiac status, respiratory status, facial features, joints).
- Initiate timely confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide family with basic information about MPS I.

Diagnostic Evaluation: Confirmatory alpha-L-iduronidase enzyme assay, urine and/or blood spot mucopolysaccharides. Patients with low alpha-L-iduronidase enzyme assay and elevated mucopolysaccharides in urine or blood should have *IDUA* gene analysis in consultation with the pediatric genetic/metabolic specialist.

Clinical Expectations: The clinical presentation and severity of symptoms of MPS I are variable, ranging from severe disease to attenuated variants (historically known as Hurler-Scheie disease and Scheie disease) that generally present with a later onset and a milder clinical presentation. In general, symptoms may include coarse facies, progressive dysostosis multiplex, hepatosplenomegaly, corneal clouding, hearing loss, mental retardation or learning difficulties, and cardiac valvular disease. MPS I is caused by mutations in the *IDUA* gene and has an estimated incidence of approximately 1 in 30,000 live births. Treatment options include hematopoietic stem cell transplantation and enzyme replacement therapy (ERT). ERT administration is highly complicated and should only be given under the guidance of a specialist with expertise in LSD.

Additional Information

Genetics Home Reference

Genetic Testing Registry

Baby's First Test

Mayo Clinic Laboratories Testing

MPSWB / Mucopolysaccharidosis, Blood

MPSBS / Mucopolysaccharidosis, Blood Spot

MPSSC / Mucopolysaccharides Screen, Random, Urine

MPS1Z / Hurler Syndrome, Full Gene Analysis, Varies

IDUAW / Alpha-L-Iduronidase, Blood