Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced Alpha-L-iduronidase (IDUA) with Elevations in Dermatan Sulfate and Heparan Sulfate

This screening result is suggestive of a biochemical diagnosis of MPS I. Molecular genetic testing is pending and MDH will provide the results to the metabolic specialist seeing your patient.

Next Steps

This week, you should take the following recommended actions:

- **Consult** with metabolic specialist. Contact information for the metabolic specialists can be found on the newborn screening report and on the resource list provided.
- **Contact** family to notify them of the newborn screening result and assess symptoms as guided by the metabolic specialist.
- **Arrange** referral to metabolic specialist for a comprehensive evaluation.

If you have questions about the newborn screening result or your next steps, an on-call Newborn Screening Program genetic counselor is available at (651) 201-3548.

Review with Family

Discuss this result with the family as MDH has **not** notified them. Share your follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns.

Differential Diagnosis

Absent/reduced IDUA with elevations in dermatan sulfate and heparan sulfate are primarily associated with:

- Mucopolysaccharidosis type 1 (MPS I) — Incidence of 1 in 100,000

Clinical Summary

MPS I is a lysosomal disorder caused by a deficiency in the enzyme, alpha-L-iduronidase (IDUA). As a result of this deficiency, glycosaminoglycans (i.e., dermatan sulfate and heparan sulfate) accumulate, leading to the signs and symptoms of MPS I.

MPS I is divided into two forms: severe and attenuated. Severe MPS I is associated with multi-system involvement, including progressive and rapid neurocognitive impairment. Symptoms usually appear in the first or second year of life. In the attenuated form, onset occurs between three years of age and adulthood with slower progression than the severe form. Central nervous system (CNS) involvement is not typically a component of the attenuated form. Some individuals have low enzyme activity, but do not develop disease—referred to as “pseudodeficiency.”

Enzyme replacement therapy (ERT) is available and has been shown to slow or stabilize disease progression. The primary treatment for the majority of children with the severe form is hematopoietic stem cell transplantation.