Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced Alpha-L-iduronidase (IDUA) with Elevated MPS I Biomarker

Next Steps

This week, you should take the following recommended actions:

- **Consult** with a metabolic specialist. Contact information for the metabolic specialists can be found on the resource list provided or our website.
- **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 elevated MPS I biomarker (endogenous MPS I specific glycosaminoglycan fragment) is 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, complex sugar molecules called glycosaminoglycans (GAGs) accumulate, leading to the variable signs and symptoms of MPS I.

MPS I is divided into two forms: severe and attenuated. Severe MPS I, sometimes called Hurler syndrome, 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 typically 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.”

Treatment

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. Supportive therapies and management like surgery and physical therapy can also be beneficial.

Individuals with MPS I should follow with a metabolic specialist and their regular doctor for continued monitoring of development.

DEPARTMENT OF HEALTH

Minnesota Newborn Screening Program | www.health.state.mn.us/newbornscreening
P.O. Box 64899, St. Paul, MN 55164
To obtain this information in a different format:
contact health.newbornscreening@state.mn.us
or call (800) 664-7772 ***interpreters available***

REV 4/2026