Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Absent/Reduced Biotinidase (BTD)

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.
- **Contact** family to notify them of the newborn screening result and assess symptoms.
- **Arrange** laboratory testing as recommended by the metabolic specialist.

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 the follow-up plan with them. Educate family about signs, symptoms, and when to contact you with concerns.

False Positives

Screening result can be impacted by transfusion and environmental factors such as heat and humidity.

Differential Diagnosis

Absent/reduced biotinidase is primarily associated with:

- Biotinidase deficiency — Incidence of 1 in 50,000 (more common in Somali populations)

Clinical Summary

Biotinidase deficiency is caused by a deficiency in the enzyme, BTD. As a result of this deficiency, the body is unable to process the vitamin, biotin. This can lead to a buildup of potentially toxic compounds. There are two forms of biotinidase deficiency: profound and partial.

Profound biotinidase deficiency can cause seizures, hypotonia, breathing problems, hearing and vision loss, ataxia, skin rashes, alopecia, and candidiasis. Intellectual disabilities can also occur.

Partial biotinidase deficiency is a milder form of this condition. Affected children may experience hypotonia, skin rashes, and alopecia. These symptoms may only appear during illness, infection, or other times of stress.

Treatment consists of lifelong biotin supplementation. If treated early, complications of biotinidase deficiency can be prevented or improved if they have already developed.