Provider Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Significantly Elevated Psychosine

Differential Diagnosis

Infantile Krabbe disease — Incidence of 1 in 100,000

False Positives

Unlikely.

Next Steps

Today, you should take the following recommended actions:

- **Call** (612) 672-7575 to speak with the on-call metabolic specialist at M Health Fairview IMMEDIATELY for guidance on clinical follow-up. M Health Fairview is the only center in the region equipped to provide treatment for infantile Krabbe disease.
- **Contact** family to notify them of the newborn screening result as MDH has **not** notified them. Share follow-up plan with them.
- **Educate** family about signs, symptoms and when to reach out with concerns.
- **Arrange** referrals and help family coordinate follow-up.

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.

Clinical Summary

Krabbe disease is a neurometabolic disorder. It is caused by a deficiency in the GALC enzyme. The GALC enzyme is responsible for the turnover of the myelin sheath. It also breaks down psychosine, a lipid that is a byproduct of the creation of myelin. In high amounts, psychosine is toxic to the body and can lead to the deterioration of the myelin sheath and nerve damage.

There are two main types of Krabbe disease: infantile and late-onset. The infantile type is more severe and most likely to occur when psychosine levels are above 10 nmol/L. Symptoms of infantile Krabbe disease usually present within the first six weeks of life. Initial symptoms include irritability, sensitivity to loud noises, feeding difficulties, and muscle stiffness. If untreated, it can cause:

- Seizures
- Blindness
- Deafness
- Neurologic deterioration
- Death within the first two years of life

There is no cure for Krabbe disease. Currently, the only treatment available is hematopoietic stem cell transplantation (HSCT). HSCT is most effective when performed before symptoms develop. HSCT has been shown to slow the progression of the disease, improve quality of life, and increase the lifespan. Supportive therapies and management like physical therapy and medications can also be beneficial. Even when performed pre-symptomatically, treatment does not prevent all morbidity and mortality.