

# Positive Result: Blood Spot Screen Result Notification



## Elevated Psychosine

### Differential Diagnosis

Elevated psychosine is primarily associated with:

- Krabbe disease — Incidence of 1 in 250,000

Other disorders to consider:

- Saposin A deficiency — described in <10 patients

### False Positives

Unlikely

### Next Steps

This week, you should take the following recommended actions:

- **Call** a metabolic specialist for guidance on clinical follow-up within one business day. Contact information for the metabolic specialists can be found on the resource list provided.
- **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 the most severe with death by two years of age if left untreated. The late-onset type is less severe and most likely to occur when psychosine levels are between 2-10 nmol/L.

Late-onset Krabbe disease can begin as early as one year of age. Vision problems often appear first and then trouble with walking. Signs and symptoms can vary widely from one person to the next and will worsen over time.

Children with late-onset Krabbe are still at risk for leukodystrophy and benefit from early treatment. Clinical monitoring is needed to determine the best time to treat (ideally before symptoms develop).

There is no cure for Krabbe disease. Currently, the only treatment available is hematopoietic stem cell transplantation (HSCT). 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.