

Positive Result: Blood Spot Screen Result Notification



Significantly Elevated Psychosine: Risk for Krabbe Disease

What was found on the newborn screen?

The newborn screen collected at birth found your baby has low levels of an enzyme called galactosylceramidase (GALC) and high levels of a fat called psychosine.

What does this mean?

This means your baby likely has Krabbe disease, but more testing is needed.

There are two main types of Krabbe disease: infantile and late-onset. The psychosine levels found on your baby's newborn screen are suggestive of the infantile type.

What happens next?

Your baby's doctor will help arrange for more testing with specialists familiar with Krabbe disease to confirm the result. Due to the importance of early treatment, this process will move quickly. Your doctor and specialists are there to support you.

What causes Krabbe disease?

Krabbe disease happens when children are missing all or some of the GALC enzyme. GALC breaks down psychosine. When psychosine is not broken down, it builds up in the body and can lead to damage to the nerves.

What health problems can it cause?

Krabbe disease is different for each child. It is a lifelong condition that may result in serious health problems.

Initial symptoms of infantile Krabbe disease include:

- extreme irritability
- sensitivity to loud noises or touch
- feeding difficulties
- muscle weakness

If untreated, it can lead to intellectual disability, blindness, deafness, decreased mobility, seizures, and death. Children with Krabbe disease can benefit from prompt and careful treatment.

What treatment options are available?

Currently, the only treatment is a stem cell transplant (HSCT), which is best when done before symptoms start. Krabbe disease cannot be cured. HSCT can help to slow the disease, improve quality of life, and increase the lifespan. Even with early treatment, children with infantile Krabbe may still have significant developmental delays.

Supportive therapies and management like physical therapy and medications can also help.

Children with Krabbe disease should see their regular doctor and providers who specialize in Krabbe disease.

Resources



Scan here for more information about newborn screening in Minnesota or visit our website:
www.health.state.mn.us/people/newbornscreening

Newborn Screening Info Center:
www.newbornscreening.hrsa.gov

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org