NORTH CAROLINA NEWBORN SCREENING PROGRAM

Krabbe Disease Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain diseases or other health problems that can be treated if diagnosed early in life. The newborn screening result showed that your baby might have Krabbe disease. Your baby will be referred to a specialist for more testing to know for sure.

There are usually no signs of Krabbe disease at birth.

What is Krabbe disease?

Krabbe disease is a rare genetic condition that can damage the nerves. People with Krabbe disease cannot produce an enzyme called galactosylceramidase (*GALC*). *GALC* breaks down a chemical called psychosine. When *GALC* is absent or low, psychosine builds up in the body and can cause serious health problems. There are two types of Krabbe disease: infantile and late-onset. **Babies with infantile Krabbe need to be seen by a specialist as soon as possible.**

What are the symptoms of Krabbe disease?

Krabbe disease is a life-long condition and is different for each child.

Signs and symptoms of the infantile form of Krabbe disease usually start within the first few weeks to months of life. In the late-onset form of Krabbe disease, symptoms may not appear until later in infancy, childhood, adolescence, or adulthood.

Signs of the infantile form may include:

- Feeding problems (may be mistaken for colic)
- Extreme irritability
- Stiffness and muscle spasms
- Loss or failure to gain developmental milestones
- Muscle weakness
- Seizures
- Vision loss

What happens next?

Your child will be referred to Krabbe specialists at the Duke Pediatric Transplant and Cellular Therapy (PTCT) Program. They will schedule an appointment within 1–2 days. The doctors will check your baby's blood for psychosine levels and GALC enzyme activity. If the GALC enzyme is low or missing, your baby is likely to have Krabbe. Additional follow-up tests may also be done including:

- MRI of the brain
- Checking protein levels in the spinal fluid
- Nerve testing
- DNA testing for the mutation in the *GALC* gene

How is Krabbe disease treated?

It is important to talk to your health care provider about which treatment(s) are best for your baby. Treatments may include the following:

- Stem cell transplant: Bone marrow or cord blood cells from a donor are given to your baby through their bloodstream. After the transplant takes, their body can make the missing enzyme. This treatment is recommended for babies with infantile Krabbe and for children with late-onset Krabbe.
- Treatments to address symptoms like:
  - o Reflux
  - o Feeding problems
  - o Spasticity (tight muscles)
  - o Respiratory problems
- Gene therapy and other research clinical trials may also be available.

Children who receive early and ongoing treatment for Krabbe disease can live longer and have a better quality of life.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Hunters Hope

Krabbe Connect

Newborn Screening Information Center

Baby's First Test

Duke PTCT Program

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health
www.ncdhhs.gov
https://slph.dph.ncdhhs.gov/newborn/default.asp
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