TEXAS Health and Human Services
Texas Department of State Health Services
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Commissioner

Newborn Screening FACT Sheet

# Krabbe Disease

**What is Krabbe Disease?**

Krabbe Disease is a lysosomal disease that prevents the body from recycling galactolipids. Galactolipids are important for cells in the body to work properly, but when they cannot be recycled, they start to build up. High levels of galactolipids destroy the protective covering of nerve cells called myelin. The enzyme galactosylceramidase (GALC) breaks down galactolipids to help provide energy. This process takes place in lysosomes, the recycling compartments in your cells. Krabbe Disease develops when GALC enzyme activity decreases. This makes it harder for lysosomes to break down galactolipids. Two galactolipids, galactosylceramide and psychosine, build up. This buildup destroys myelin and prevents nerve signals from traveling throughout the body. This damage leads to the signs and symptoms of the condition.

It is an inherited in an autosomal recessive pattern, meaning both parents are genetic carriers of the disease but do not have the disease itself. There are two types of Krabbe Disease that differ in signs and symptoms and age of onset:

- Early infantile Krabbe Disease
- Later-onset Krabbe Disease

In the United States, Krabbe Disease affects about 1 in 100,000 individuals.

**What are the Symptoms?**

Signs of Krabbe Disease can appear in the first few months after birth, particularly in the early infantile-onset form. The later-onset form of Krabbe Disease may not appear until later in infancy, childhood, adolescence, or adulthood.

Signs of the condition may include the following:

- Irritability
- Muscle weakness
- Stiffness and muscle spasms
- Feeding problems
- Fevers
- Seizures
- Loss of motor milestones (milestone regression)
- Vision loss

**What to Expect with Krabbe Care?**

It is important to talk to your health care provider about which treatment(s) are best for your baby. The goal of treatment is to try to address the health problems caused by this condition.

The only available therapy is hematopoietic stem cell transplantation and that is most effective if performed before 30 days of life in patients with the infantile form or prior to the onset of clinical symptoms in the late-onset forms.

Treatments may include the following:

- Stem cell transplant (SCT)
- Specific treatments to address health problems like:
  - Reflux
  - Feeding problems
  - Spasticity
  - Respiratory problems
- Gene Therapy and other research clinical trials may also be available.

Children who receive early and ongoing treatment for Krabbe Disease may live longer and have fewer health issues.

**Resources:**

**Baby's First Test**

**https://www.babysfirsttest.org/newborn-screening/conditions/krabbe**

**Condition Information for Families- HRSA Newborn Screening Clearinghouse**

https://newbornscreening.hrsa.gov/conditions/krabbe-disease

**National Organization for Rare Diseases**

https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/

**University of Washington, Gene Reviews**

https://medlineplus.gov/genetics/condition/krabbe-disease/

**Navigate Life Texas**

https://www.navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed

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**Where do I go for more information?**
Use your phone's camera to scan the QR code.

# Next steps after a positive Newborn Screen for Krabbe

Newborn screening is a blood test performed on every baby that identifies those at risk for rare disorders. Your baby's results show absent or reduced B-galactocerebrosidase (GALC) activity. This means there is a chance that your baby may have a condition called Krabbe Disease. More confirmatory testing and an exam are needed as soon as possible to see if your child has Krabbe.

## What happens next?

- It is important to follow instructions provided to you to have the laboratory tests drawn. These results will help confirm or rule out the diagnosis.
- Your baby's doctor will help arrange a visit to a specialist clinic that is familiar with Krabbe. The specialist will check your baby's health and discuss the test results in more detail at the visit. The team may also include a genetic counselor because this is a genetic disease. It is important to keep this appointment. A quick diagnosis and early treatment may lessen the impact of the disease.

## What is Krabbe?

Krabbe is due to a change, also known as a mutation, in a gene that encodes for an enzyme called galactocerebroside beta-galactosidase (GALC). This change means that the body does not create GALC properly, which is necessary for the production of myelin. Myelin is a protective material that surrounds the nerves. When there is not enough GALC to create myelin, a toxin is created instead which leads to the death of brain cells and nerves. There are different types of Krabbe. The specialists will help figure out which type, if any, your child has.

## What are the symptoms of Krabbe?

The severity and age when problems begin depends on which type of Krabbe a person has. Loss or underdevelopment of motor skills, floppy muscle tone, hearing loss or sensitivity to loud noises, irritability, vision loss or failure to thrive can occur. In early onset Krabbe, problems begin between two to six months of age.

**What is the treatment?** Early treatment involves supportive care for each organ affected and management of symptoms as needed. Treatment can include Enzyme Replacement Therapy (ERT) or stem cell transplant for those with more severe symptoms.

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