

Information Sheet

What is Krabbe Disease?

Krabbe disease (also called globoid cell leukodystrophy) is a genetic condition that affects the body's nervous system. It is part of a group of rare conditions called lysosomal storage disorders and leukodystrophies. This means the body is missing an important enzyme (a type of protein) needed to protect the nerves in the brain and spinal cord. Without it, the protective coating around the nerves, called myelin, breaks down. If left untreated, Krabbe disease can cause severe muscle stiffness, loss of vision or hearing, and a shortened lifespan.

Krabbe disease can be found early through newborn screening. If a baby's screen shows a risk for Krabbe, more testing is done using a blood sample to confirm it. A regular doctor or a metabolic specialist will order these extra tests and help coordinate your baby's care. A specialist can also help arrange quick treatment if it is needed.

**About 1 in
100,000**

births in the US
are affected by
Krabbe disease.

Krabbe is treatable.

Early diagnosis through newborn screening is very important. For the severe form of the disease, treatment should start before symptoms begin to help protect the brain and improve long-term health outcomes.

Symptoms

There are different types of Krabbe disease based on when symptoms start (early infantile, late infantile, juvenile, and adult). The early infantile type is the most common and the most severe. Babies who show symptoms very early in life tend to get sick faster than those who develop symptoms later in childhood or as adults.

Common symptoms of infantile Krabbe disease include:

- Extreme fussiness and crying
- Stiff muscles or muscle spasms
- Feeding difficulties and trouble gaining weight
- Loss of skills (like no longer smiling, reaching, or sitting up)
- Unexplained fevers
- Seizure

Treatment

There is currently no cure for Krabbe disease, but there are treatments that can help. Early intervention is critical. For the best results, a stem cell transplant or bone marrow transplant must be done in the first few weeks of life, before symptoms start. This can help slow the disease and give the child a better quality of life.

Treatment also includes a team approach to help manage symptoms and keep the child comfortable. This can include physical therapy, occupational therapy, medications to relax muscles, and feeding support.

If left untreated, the severe early form of Krabbe disease causes life-threatening breathing, feeding, and nerve problems, and death.

Act Quickly

Connect with a specialist when an abnormal screening result is received.

Support & Resources

Additional Krabbe Disease Resources:

- **Hunter's Hope Foundation:** <https://www.huntershope.org/>
- **KrabbeConnect:** <https://krabbeconnect.org/>
- **United Leukodystrophy Foundation:** <https://ulf.org/>
- **National Organization for Rare Disorders (NORD):** <https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/>

Additional Newborn Screening Information:

- **Baby's First Test:** <https://www.babysfirsttest.org/newborn-screening/conditions/krabbe>
- **Gene Reviews:** <https://www.ncbi.nlm.nih.gov/books/NBK1238/>
- **GARD (Genetic and Rare Disease Information Center):** <https://rarediseases.info.nih.gov/diseases/20343/infantile-krabbe-disease>
- **Medline Plus Condition Information for Families:** <https://medlineplus.gov/genetics/condition/krabbe-disease/>
- **HRSA Newborn Screening Clearinghouse:** <https://newbornscreening.hrsa.gov/conditions/krabbe-disease>

Kansas Department of Health and Environment Resources:

- **Kansas Newborn Screening Program:** Phone: 785-296-3363; Fax: 785-559-4245 (for reporting purposes)
- **Kansas Special Health Care Needs Program:** Care Coordination and resource assistance <https://nbss.ks.gov/shcn/>