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COMMENTARY

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# Signs and Symptoms of Krabbe Disease

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## Description

Krabbe disease is a lysosomal storage disorder that is uncommon and frequently fatal and causes gradual harm to the nervous system. KD includes abnormal sphingolipid metabolism and has an autosomal recessive inheritance pattern.

### Signs and symptoms

Asymptomatic infantile-onset Krabbe disease (12 months after birth) and later-onset Krabbe disease have different symptom patterns. 85–90% of people with infantile-onset Krabbe disease show progressive neurologic decline in infancy and pass away before the age of two. Angry behaviour, fevers, limb stiffness, seizures, eating issues, vomiting, staring spells, and a delay of mental and motor development are among the symptoms. Doctors sometimes confuse the disease's early signs for those of cerebral palsy. Other signs and symptoms include stiffness, deafness, optic atrophy, enlargement of the optic nerve, blindness, paralysis, and difficulties swallowing. Long-term weight loss is also possible. 15% to 20% of people with Krabbe disease who develop it later experience a substantially slower course of the disease. These people may also exhibit symptoms like esotropia, slurred speech, and sluggish or absent motor milestone development.

**Causes:** A mutation in the autosomal recessively inherited GALC gene, which is found on chromosome 14, results in Krabbe disease. Galactosylceramidase deficiency is brought on by mutations in the GALC gene. Rarely, it could be brought on by a shortage of active saposin A. Unmetabolized lipid accumulation impairs the development of the myelin sheath that protects nerves, causing demyelination and a severe and progressive decline in motor function. Krabbe disease, which belongs to a class of conditions known as leukodystrophies, is brought on by the improper growth and development of myelin. Additionally, a lack of galactosylceramidase leads to the accumulation of psychosine, a glycosphingolipid that is toxic to oligodendrocytes, a class of non-neuronal cells referred to as neuroglia in the nervous system.

**Diagnosis:** There are several methods for determining whether Krabbe disease is present. Dried blood cell activity testing for the GALC enzyme and molecular testing for the presence of GALC enzyme mutations are both used in the newborn screening for Krabbe illness. Referrals for additional diagnostic tests and neurological examinations should be made for infants exhibiting poor enzyme activity and/or enzyme mutations. All Krabbe disease patients who exhibit symptoms have 0–5% GALC enzyme activity. A Krabbe disease signature might be found in dried blood spots with a high concentration of psychosine. The disease's unique clustering of certain cells, nerve demyelination and degeneration, and brain cell death can all be used to diagnose it. The diagnosis may be aided by the use of special myelin stains.

**Treatment:** Bone marrow or hematopoietic stem cell transplantation has been demonstrated to help cases early in the course of the disease, despite the fact that there is no known treatment for Krabbe disease. The disorder is often treated with supportive and symptomatic care. Physical therapy may support or improve circulation and muscular tone. According to a 15-year study on the developmental outcomes of Krabbe disease patients who underwent HSCT in the first seven weeks after birth, patients have a better outlook on their lifespan and functional abilities, and the disease advances more slowly. If detected early enough, even symptomatic Krabbe disease sufferers may benefit from HSCT. Usually, the stem cells used in transplants come from umbilical cord blood. Patient enrollment in gene therapy clinical studies is presently underway.

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