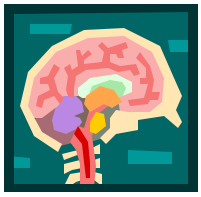

Disability Awareness Begins With You:

Krabbe Disease

What is Krabbe disease?

Krabbe disease is a degenerative disorder that affects the nervous system. It is caused by the shortage (deficiency) of an enzyme called galactosylceramidase (GALC). This enzyme deficiency results in defective myelin, the covering that insulates many nerves. Krabbe disease is considered part of a group of disorders known as leukodystrophies, which result from the imperfect growth and development of myelin.

Krabbe disease usually begins before the age of 1 year (early-onset form). Initial signs and symptoms often include feeding difficulties, episodes of unexplained fever, stiff posture, and developmental delay. As the disease progresses, muscles continue to weaken, affecting the infant's ability to move, chew, swallow, and breathe. Affected infants also experience vision loss and mental retardation. Less commonly, onset can occur in later in childhood, adolescence, or adulthood (late-onset form).

How common is Krabbe disease?

Worldwide, Krabbe disease occurs in about 1 in 100,000 to 200,000 births. A higher incidence (6 cases per 1,000 live births) has been reported in a few isolated communities in Israel.

What genes are related to Krabbe disease?

Mutations in the GALC gene cause Krabbe disease.

Mutations in the GALC gene cause a deficiency of the enzyme galactosylceramidase. This deficiency leads to a progressive loss of myelin that covers many nerves. Without myelin, nerves in the brain and other parts of the body cannot function properly, leading to the signs and symptoms of Krabbe disease.

How do people inherit Krabbe disease?

This condition is inherited in an autosomal recessive pattern, which means two copies of the gene in each cell are altered. Parents of an individual with this autosomal recessive disorder are carriers of one copy of the altered gene but do not show signs and symptoms of the disorder.

Where can I find additional information about Krabbe disease?

You may find the following resources about Krabbe disease helpful.**

- NIH Publications - National Institutes of Health (3 links)
- MedlinePlus - Health Information (2 links)
- Educational resources - Information pages (5 links)
- Patient support - For patients and families (4 links)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.**

- Gene Reviews - Clinical summary
- Gene Tests - DNA tests ordered by healthcare professionals
- ClinicalTrials.gov - Linking patients to medical research
- PubMed - Recent literature
- OMIM - Genetic disorder catalog

What other names do people use for Krabbe disease?

- Diffuse Globoid Body Sclerosis
- Galactosylceramidase Deficiency Disease
- Galactosylceramide lipidosis
- galactosylcerebrosidase deficiency
- galactosylsphingosine lipidosis
- GALC deficiency
- globoid cell leukodystrophy (GCL, GLD)
- globoid cell leukoencephalopathy
- late-onset Krabbe disease (LOKD)
- psychosine lipidosis

**Visit the website (noted below) for access to these links.

From:
http://ghr.nlm.nih.gov/condition=krabbedisease