



## Krabbe disease

Krabbe disease (also called globoid cell leukodystrophy) is a degenerative disorder that affects the nervous system. It is caused by the shortage (deficiency) of an enzyme called galactosylceramidase. This enzyme deficiency impairs the growth and maintenance of myelin, the protective covering around certain nerve cells that ensures the rapid transmission of nerve impulses. Krabbe disease is part of a group of disorders known as leukodystrophies, which result from the loss of myelin (demyelination). This disorder is also characterized by the abnormal presence of globoid cells, which are globe-shaped cells that usually have more than one nucleus.

The symptoms of Krabbe disease usually begin before the age of 1 year (the infantile form). Initial signs and symptoms typically include irritability, muscle weakness, feeding difficulties, episodes of fever without any sign of infection, stiff posture, and slowed mental and physical development. 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 seizures.

Less commonly, onset of Krabbe disease can occur in childhood, adolescence, or adulthood (late-onset forms). Visual problems and walking difficulties are the most common initial symptoms in this form of the disorder, however, signs and symptoms vary considerably among affected individuals.

### Frequency

In the United States, Krabbe disease affects about 1 in 100,000 individuals. A higher incidence (6 cases per 1,000 people) has been reported in a few isolated communities in Israel.

### Genetic Changes

Mutations in the *GALC* gene cause Krabbe disease. These mutations 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.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Other Names for This Condition

- Diffuse Globoid Body Sclerosis
- Galactosylceramidase Deficiency Disease
- Galactosylceramide lipidosis
- galactosylcerebrosidase deficiency
- galactosylsphingosine lipidosis
- GALC deficiency
- GCL
- GLD
- psychosine lipidosis

## Diagnosis & Management

### Genetic Testing

- Genetic Testing Registry: Galactosylceramide beta-galactosidase deficiency  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0023521/>

### Other Diagnosis and Management Resources

- Baby's First Test  
<http://www.babysfirsttest.org/newborn-screening/conditions/krabbe>
- GeneReview: Krabbe Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1238>
- MedlinePlus Encyclopedia: Krabbe disease  
<https://medlineplus.gov/ency/article/001198.htm>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Krabbe disease  
<https://medlineplus.gov/ency/article/001198.htm>
- Health Topic: Leukodystrophies  
<https://medlineplus.gov/leukodystrophies.html>
- Health Topic: Newborn Screening  
<https://medlineplus.gov/newbornscreening.html>

### Genetic and Rare Diseases Information Center

- Krabbe disease  
<https://rarediseases.info.nih.gov/diseases/6844/krabbe-disease>

### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Krabbe Disease  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Krabbe-Disease-Information-Page>
- National Institute of Neurological Disorders and Stroke: Leukodystrophy  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Leukodystrophy-Information-Page>
- National Institute of Neurological Disorders and Stroke: Lipid Storage Diseases  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Lipid-storage-diseases-Information-Page>

### Educational Resources

- MalaCards: krabbe disease  
[http://www.malacards.org/card/krabbe\\_disease](http://www.malacards.org/card/krabbe_disease)
- Orphanet: Krabbe disease  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=487](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=487)
- Tulane University  
<http://www2.tulane.edu/tnprc/diseases/krabbe/>

## Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Diseases  
<http://www.climb.org.uk>
- Hunter's Hope Foundation  
<http://huntershope.org/>
- National Organization for Rare Disorders  
<https://rarediseases.org/rare-diseases/leukodystrophy-krabbes/>

## GeneReviews

- Krabbe Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1238>

## ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22krabbe+disease%22>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Leukodystrophy,+Globoid+Cell%5BMAJR%5D%29+AND+%28Krabbe+disease%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- KRABBE DISEASE  
<http://omim.org/entry/245200>

## **Sources for This Summary**

- GeneReview: Krabbe Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1238>
- National Institute of Neurological Disorders and Stroke: Krabbe Disease  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Krabbe-Disease-Information-Page>
- Pastores GM. Krabbe disease: an overview. *Int J Clin Pharmacol Ther.* 2009;47 Suppl 1:S75-81. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20040316>
- Sakai N. Pathogenesis of leukodystrophy for Krabbe disease: molecular mechanism and clinical treatment. *Brain Dev.* 2009 Aug;31(7):485-7. doi: 10.1016/j.braindev.2009.03.001. Epub 2009 Mar 29. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19332366>

- Suzuki K. Globoid cell leukodystrophy (Krabbe's disease): update. *J Child Neurol.* 2003 Sep;18(9): 595-603. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14572137>
  - Wenger DA, Rafi MA, Luzi P, Datto J, Costantino-Ceccarini E. Krabbe disease: genetic aspects and progress toward therapy. *Mol Genet Metab.* 2000 May;70(1):1-9. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/10833326>
- 

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/condition/krabbe-disease>

Reviewed: August 2012

Published: September 12, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services