



## GALC gene

galactosylceramidase

### Normal Function

The *GALC* gene provides instructions for making an enzyme called galactosylceramidase. Through a process called hydrolysis, this enzyme uses water molecules to break down certain fats called galactolipids, which are found primarily in the nervous system and kidneys.

Within cells, galactosylceramidase is found in enzyme-filled sacs called lysosomes where it hydrolyzes specific galactolipids, including galactosylceramide and psychosine. Galactosylceramide is an important component of myelin, the protective covering around certain nerve cells that ensures the rapid transmission of nerve impulses. Its breakdown by galactosylceramidase is part of the normal turnover of myelin that occurs throughout life. Psychosine, which is toxic to cells, forms during the production of myelin and is quickly broken down by galactosylceramidase. Under normal conditions, tissues contain very little psychosine.

### Health Conditions Related to Genetic Changes

#### Krabbe disease

More than 200 *GALC* gene mutations that cause Krabbe disease have been identified. Krabbe disease is a brain disorder that usually begins in infancy (infantile Krabbe disease) and causes movement and eating problems, impaired development, and seizures. The most common mutation in affected individuals of European ancestry (often called 30-kb del) deletes a large segment of the *GALC* gene. Other mutations insert additional DNA building blocks (nucleotides) into the *GALC* gene, delete a small number of nucleotides from the gene, or replace single nucleotides with incorrect nucleotides.

These *GALC* gene mutations severely reduce or eliminate the activity of the galactosylceramidase enzyme. As a result, galactosylceramide and psychosine cannot be broken down. The accumulation of these galactolipids causes damage to myelin-forming cells, which impairs the formation of myelin and leads to the loss of myelin (demyelination) in the nervous system. Without myelin, nerves in the brain and other parts of the body cannot transmit signals properly, leading to the signs and symptoms of Krabbe disease.

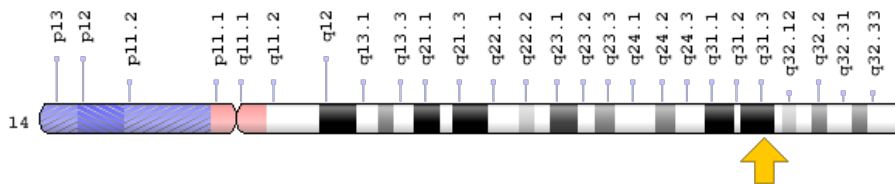
Some individuals develop symptoms of Krabbe disease in childhood, adolescence, or adulthood (late-onset Krabbe disease). It is thought that these individuals have a mutation that allows some activity of the galactosylceramidase enzyme, which

delays onset of the condition. The severity of the condition may also be affected by the presence of additional common variations (polymorphisms) in the *GALC* gene that affect the activity of the galactosylceramidase enzyme.

## Chromosomal Location

Cytogenetic Location: 14q31.3, which is the long (q) arm of chromosome 14 at position 31.3

Molecular Location: base pairs 87,933,014 to 87,993,665 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- galactocerebrosidase
- galactosylceramide beta-galactosidase
- GALC\_HUMAN
- GALCERase

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (sixth edition, 1999): Synthesis and Metabolism of Myelin  
<https://www.ncbi.nlm.nih.gov/books/NBK28068/>

### GeneReviews

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

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GALC%5BTIAB%5D%29+OR+%28galactosylceramidase%5BTIAB%5D%29%29+OR+%28%28galactocerebrosidase%5BTIAB%5D%29+OR+%28Galactosylceramide+beta-Galactosidase%5BTIAB%5D%29+OR+%28GALCERase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

## OMIM

- GALACTOSYLCERAMIDASE  
<http://omim.org/entry/606890>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GALC.html](http://atlasgeneticsoncology.org/Genes/GC_GALC.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GALC%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4115](https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4115)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2581>
- UniProt  
<http://www.uniprot.org/uniprot/P54803>

## **Sources for This Summary**

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Reviewed: January 2018  
Published: February 20, 2018

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
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