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# Galactosemia

Información en español (/espanol/

**Categories:** Congenital and Genetic Diseases (/diseases/diseases-by-category/5); Newborn Scr
(/diseases/diseases-by-category/37)

**Subtypes:** Classic galactosemia (/diseases/13639/classic-galactosemia); Duarte Galactosemia
(/diseases/12908/duarte-galactosemia); Galactokinase deficiency (/diseases/2422/galactokinas
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## Summary

**Galactosemia**, which means “galactose in the blood,” refers to a group of inherited disorder
body's ability to process and produce energy from a sugar called galactose. When people wit
injest foods or liquids containing galactose, undigested sugars build up in the blood. Galacto
many foods, including all dairy products (milk and anything made from milk), many baby forn
fruits and vegetables.[1][2] The impaired ability to process galactose can be due to the deficie
enzymes, caused by mutations in different genes.[3] There are 3 main types of galactosemia
distinguished based on their genetic causes, signs and symptoms, and severity:[1][3][4][5][6]

**Classic galactosemia (type 1)** (https://www.babysfirsttest.org/newborn-screening/cor
galactosemia) - the most common and severe type, caused by mutations in the *GALT (https://ghr.nlm.nih.gov/gene/GALT)* gene, and characterized by a complete deficiency of
galactose-1-phosphate uridyl transferase (GALT). Early signs and symptoms include liver d
susceptibility to infections, failure to thrive, and cataracts. These can usually be prevented
early diagnosis and treatment, but other progressive or long-term problems are common d
These include intellectual deficits, movement disorders, and premature ovarian failure (in

**Galactokinase deficiency (type 2) (https://rarediseases.info.nih.gov/diseases/2422/galactokinase-deficiency)** - ca
mutations in the *GALK1 (https://ghr.nlm.nih.gov/gene/GALK1)* gene and characterized by
enzyme galactokinase 1. This type typically causes only the development of cataracts
(https://nei.nih.gov/health/cataract/cataract_facts), which may be prevented or resolved v
Rarely, this type causes pseudotumor cerebri (https://www.ninds.nih.gov/Disorders/All-
Disorders/Pseudotumor-Cerebri-Information-Page) (a condition which mimics the sympto
tumor when no brain tumor is present).

**Galactose epimerase deficiency (type 3) (https://www.babysfirsttest.org/newbo
screening/conditions/galactoepimerase-deficiency)** - caused by mutations in the *GA
*(https://ghr.nlm.nih.gov/gene/GALE)* gene and characterized by a deficiency of the enzym
4-epimerase. Symptoms and severity of this type depend on whether the deficiency is con
types of blood cells or is present in all tissues. Some people with this type have no signs d
others have symptoms similar to those with classic galactosemia. Like in classic galactos
symptoms can be prevented or improved with treatment.

There is also a "variant" of classic galactosemia called Duarte variant galactosemia (https://rarediseases.info.nih.gov/diseases/12908/duarte-galactosemia), in which a person h the *GALT* gene but has only partial deficiency of the enzyme. Infants with this form may have resolves when switched to a low-galactose formula. Some studies have found that people wi increased risk for mild neurodevelopmental problems, but other studies have found there is n The risk may depend on the extent of the deficiency.[3][7]

Inheritance of all types of galactosemia is autosomal recessive.[1][3] The diagnosis may be su symptoms or results of newborn screening tests, and can be confirmed by measuring enzym genetic testing.[3] Depending on the type of galactosemia, treatment may involve removing ga diet (as soon as the disorder is suspected), calcium supplementation, and individualized care symptoms.[6] The long-term outlook for people with galactosemia varies depending on the ty present, and commitment to the diet.[6]

Last up

## Symptoms

This table lists symptoms that people with this disease may have. For most diseases, sympt person to person. People with the same disease may not have all the symptoms listed. This i from a database called the Human Phenotype Ontology (HPO) (http://www.human-phenoty The HPO collects information on symptoms that have been described in medical resources. updated regularly. Use the HPO ID to access more in-depth information about a symptom.

Showing 1-5 of 47 | View All

| Medical Terms | Other Names |
|---|---|
| **80%-99% of people have these symptoms** | |
| Cognitive impairment | Abnormality of cognition [ more ] |
| Failure to thrive in infancy | Faltering weight in infancy [ more ] |
| | |

| | |
|---|---|
| Feeding difficulties in infancy | |
| Global developmental delay | |
| Hepatic failure | Liver failure |

Showing 1-5 of 47 | View All

***Do you have more information about symptoms of this disease? We want to hear (/Feedback?diseaseId=2424)***

Last u

***Do you have updated information on this disease? We want to hear from you. (/Fe diseaseId=2424)***

## Diagnosis

Making a diagnosis for a genetic or rare disease can often be challenging. Healthcare profes look at a person's medical history, symptoms, physical exam, and laboratory test results in or diagnosis. The following resources provide information relating to diagnosis and testing for t you have questions about getting a diagnosis, you should contact a healthcare professional.

### Testing Resources

The Genetic Testing Registry (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0016952/) (C information about the genetic tests for this condition. The intended audience for the GTR i providers and researchers. Patients and consumers with specific questions about a genet contact a health care provider or a genetics professional.

### Newborn Screening

The Newborn Screening Coding and Terminology Guide (http://newbornscreeningcodes.nlm.nih.gov/nb/sc/condition/GALT ) has information on th used for newborn screening tests. Using these standards helps compare data across diffe This resource was created by the National Library of Medicine.

## Find a Specialist

If you need medical advice, you can look for doctors or other healthcare professionals who ha with this disease. You may find these specialists through advocacy organizations, clinical tria published in medical journals. You may also want to contact a university or tertiary medical c because these centers tend to see more complex cases and have the latest technology and t

If you can't find a specialist in your local area, try contacting national or international specialis able to refer you to someone they know through conferences or research efforts. Some spec willing to consult with you or your local doctors over the phone or by email if you can't travel t

You can find more tips in our guide, How to Find a Disease Specialist (/guides/pages/25/how specialist). We also encourage you to explore the rest of this page to find resources that can specialists.

### Healthcare Resources

To find a medical professional who specializes in genetics, you can ask your doctor for a r search for one yourself. Online directories are provided by the American College of Medica (http://www.acmg.net/ACMG/Genetic_Services_Directory_Search.aspx) and the National S Counselors (https://www.findageneticcounselor.com/). If you need additional help, contac Information Specialist (https://rarediseases.info.nih.gov/about-gard/contact-gard). You ca about genetic consultations (https://ghr.nlm.nih.gov/primer#consult) from Genetics Home

## Research

Research helps us better understand diseases and can lead to advances in diagnosis and tre section provides resources to help you learn about medical research and ways to get involve

### Clinical Research Resources

ClinicalTrials.gov (http://www.clinicaltrials.gov/ct2/results?cond=%22Galactosemia%22) l related to Galactosemia. Click on the link to go to ClinicalTrials.gov to read descriptions of

***Please note:*** *Studies listed on the ClinicalTrials.gov website are listed for informational p being listed does not reflect an endorsement by GARD or the NIH. We strongly recommen a trusted healthcare provider before choosing to participate in any clinical study.*

Orphanet (http://www.orpha.net/consor/cgi-bin/ResearchTrials_ResearchProjects_Simple lng=EN&LnkId=355&Typ=Pat&fdp=y&from=rightMenu) lists European clinical trials, resear patient registries enrolling people with this condition.

## Organizations

Support and advocacy groups can help you connect with other patients and families, and the valuable services. Many develop patient-centered information and are the driving force behin better treatments and possible cures. They can direct you to research, resources, and service organizations also have experts who serve as medical advisors or provide lists of doctors/cli group's website or contact them to learn about the services they offer. Inclusion on this list is endorsement by GARD.

### Organizations Supporting this Disease

Galactosemia Foundation (/organizations/606)
P.O. Box 1512
Deerfield Beach, FL 33443
Toll-free: 866-900-7421
E-mail: outreach@galactosemia.org (mailto:outreach@galactosemia.org)
Website: http://www.galactosemia.org (http://www.galactosemia.org)

Metabolic Support UK (/organizations/138)
5 Hilliards Court
Sandpiper Way
Chester Business Park
Chester, CH4 9QP United Kingdom
Toll-free: 0800 652 3181
Telephone: 0845 241 2173
E-mail: https://www.metabolicsupportuk.org/contact-us (https://www.metabolicsupportuk
Website: https://www.metabolicsupportuk.org (https://www.metabolicsupportuk.org)

***Do you know of an organization? We want to hear from you. (/Feedback?diseaseI***

## Living With

Living with a genetic or rare disease can impact the daily lives of patients and families. These help families navigate various aspects of living with a rare disease.

### Education Resources

The Genetics Education Materials for School Success (GEMSS) (http://www.gemssforschools.org/conditions/galactosemia/default) aims to assure that a genetic health conditions succeed in school-life. Their Web site offers general and conditi education resources to help teachers and parents better understand the needs of students conditions.

## Learn More

These resources provide more information about this condition or associated symptoms. Th resources contain medical and scientific language that may be hard to understand. You may these resources with a medical professional.

### Where to Start

Genetics Home Reference (GHR) (http://www.ghr.nlm.nih.gov/condition=galactosemia) co information on Galactosemia. This website is maintained by the National Library of Medic

MedlinePlus (http://www.nlm.nih.gov/medlineplus/ency/article/000366.htm) was designe Library of Medicine to help you research your health questions, and it provides more inforr topic.

The National Organization for Rare Disorders (http://rarediseases.org/rare-diseases/galac has a report for patients and families about this condition. NORD is a patient advocacy org individuals with rare diseases and the organizations that serve them.

The Screening, Technology And Research in Genetics (STAR-G) Project (https://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html) has a fa condition, which was written specifically for families that have received a diagnosis as a re screening. This fact sheet provides general information about the condition and answers c of particular concern to parents.

### In-Depth Information

GeneReviews provides current, expert-authored, peer-reviewed, full-text articles describing genetic testing to the diagnosis, management, and genetic counseling of patients with spe conditions.
Classic Galactosemia and Clinical Variant Galactosemia (https://www.ncbi.nlm.nih.gov/bo
Epimerase deficiency galactosemia (https://www.ncbi.nlm.nih.gov/books/NBK51671/)
Duarte Variant Galactosemia (https://www.ncbi.nlm.nih.gov/books/NBK258640/)

The Monarch Initiative (https://monarchinitiative.org/disease/OMIM:230400) brings toget condition from humans and other species to help physicians and biomedical researchers. are designed to make it easier to compare the signs and symptoms (phenotypes) of differ discover common features. This initiative is a collaboration between several academic ins

the world and is funded by the National Institutes of Health. Visit the website to explore th condition.

Orphanet (http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=352) is a Eur portal for information on rare diseases and orphan drugs. Access to this database is free

PubMed (http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=omim&cmd=Display&dopt=omim_pubmed_calculated&from_uid=230400) is a searcha medical literature and lists journal articles that discuss Galactosemia. Click on the link to search on this topic.

## News & Events

### News

NCATS Rare Diseases Are Not Rare! Challenge (/news/633)
October 9, 2018

The NIH Undiagnosed Diseases Network Expands (/news/632)
September 26, 2018

## GARD Answers

Questions sent to GARD may be posted here if the information could be helpful to others. We identifying information when posting a question to protect your privacy. If you do not want yo posted, please let us know. Submit a new question (/about-gard/contact-gard)

My 11 year-old grandson has galactosemia. He drinks soy milk all the time. Is one of the s drinking so much soy estrogen or some hormone that can cause breast development? I ha find an answer to that question. Any information you can provide will be appreciated. Thar (/diseases/2424/galactosemia/cases/56074)

***Have a question? Contact a GARD Information Specialist. (/about-gard/contact-ga***

## References

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***Do you know of a review article? We want to hear from you. (/Feedback?diseaseId***