Galactosemia Foundation
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Galactosemia Toolkit

FOR NEWLY DIAGNOSED FAMILIES

The Galactosemia Foundation's informative guide gives you the tools and resources you need to be the best advocate for your child.

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Chances are, you are feeling shocked, scared, and overwhelmed by your child's galactosemia diagnosis. Last week, you and your child's pediatrician may have never even heard of galactosemia. Now, all you think about is galactosemia. We've got you.

The Galactosemia Foundation is fueled by volunteers who once felt exactly like you do at this moment. It's natural to have a lot of questions and concerns, to worry about how you will keep up with the latest dietary guidelines, to wonder how you will adapt as a family to living with galactosemia. This toolkit is designed with all of that in mind, to help you advocate for your child.

We are here to help you learn about what you can expect as your child grows so you can better navigate challenges that may come your way. With these tools and resources at your fingertips, you can spend less time researching (sometimes scary) information on the Internet and spend more time with your beloved child.

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What Should You Expect After Diagnosis?

If your pediatrician hasn't heard of galactosemia, don't be surprised. You will be referred to a metabolic geneticist who will be familiar with the disease. While appointments may vary, you will meet with several different people during your visit, including a dietician and, likely, a social worker.

As a parent with a child diagnosed with a condition you are probably unfamiliar with, we recommend bringing a pen and paper to take notes. You may also want to have a loved one accompany you as the information presented during your initial appointment can be overwhelming. Your new team will do an assessment of your child and give an overview of what you can expect for future appointments and testing, which will vary based on age.

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Did You Know?

Galactosemia is now included as **part of routine newborn screening** in all 50 U.S. states.

> Click here to learn more about the NBS process in your state.

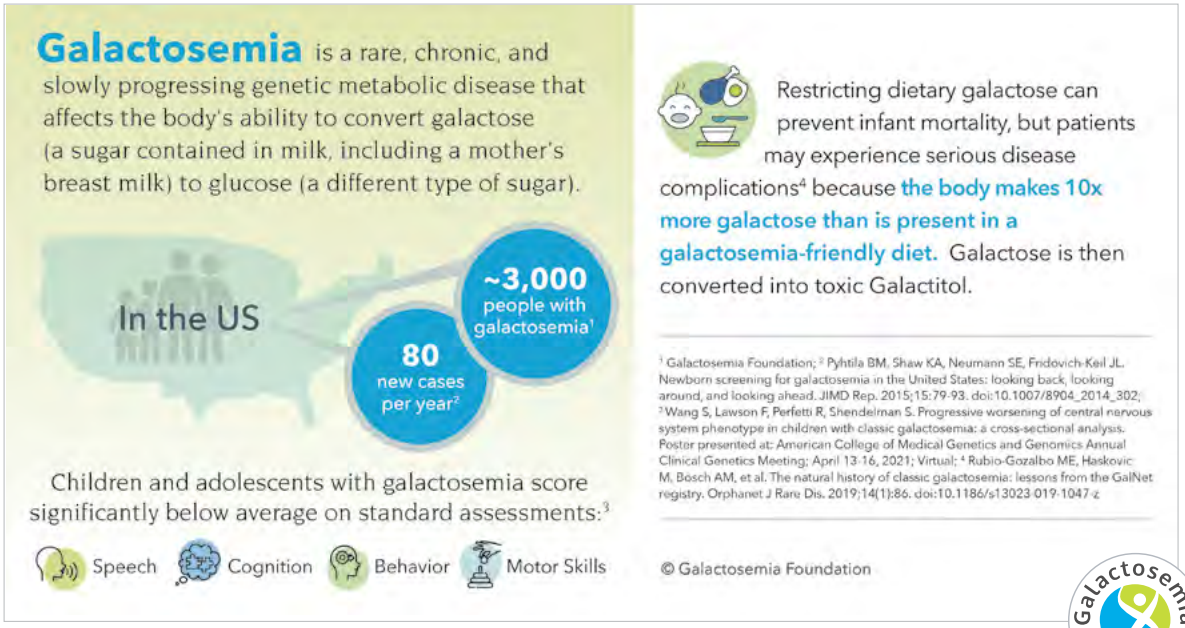
Is Your Child Newly Diagnosed with Galactosemia?

What is Galactosemia?

When you first heard the word “galactosemia,” you likely didn’t recognize it. Maybe you even had trouble pronouncing it. We understand. Galactosemia is not a highly recognized disease—though, together, we are working hard to change that—because it is rare. Galactosemia is considered an orphan disease, affecting 1 in 30,000-70,000 people.

The term “galactosemia” literally means too much galactose in the blood. It’s a hereditary disorder that affects the body’s ability to convert galactose (a sugar contained in milk, including a mother’s milk) to glucose (a different type of sugar) in the metabolic pathway. The disorder is caused by a deficiency of an enzyme called galactose-1-phosphate uridylyltransferase (GALT), which is vital to this process. The GALT enzyme deficiency leads to an abnormal accumulation of galactose-related chemicals in various organs of the body, causing the physical signs and symptoms of galactosemia (described later in this toolkit).

In galactosemia, galactose is mistakenly turned into a substance called galactitol. Galactitol is highly toxic, which means it’s harmful to the body. Toxic galactitol can build up in the blood, tissues, and organs, including the brain. There is evidence that toxic galactitol is responsible for a range of health issues that people with galactosemia may experience. There are currently no approved therapies for galactosemia, though clinical trials for a possible treatment are underway.



Click here to learn more about the science behind galactosemia.

Types of Galactosemia

Three types of galactosemia have been identified. They are caused by a mutation or deletion in the GALT, GALK1, and GALE genes, the ones responsible for making the enzymes that are essential to metabolize galactose.

Type I: Classic Galactosemia

Classic galactosemia (type I) is the most severe form. If infants with classic galactosemia are not treated quickly with a galactose-free diet, life-threatening complications occur shortly after birth.

Both parents must pass on a defective copy of the GALT gene for their child to develop classic galactosemia. The newborn screening test in all 50 states screen for this type of galactosemia.

- **Variant Duarte Galactosemia**

  Like classic galactosemia, duarte galactosemia is caused by a mutation in the GALT gene, but it is much less severe. Children with duarte galactosemia typically do not have serious symptoms and may or may not require a restricted diet.

Type II: Galactokinase Deficiency Galactosemia

Galactokinase deficiency galactosemia is due to a mutation in the GALK1 gene, making it impossible for an affected child to fully break down galactose. Not all newborn galactosemia screening tests cover this version of the deficiency, so a normal result can be misleading.

Type III: Epimerase-Deficiency Galactosemia

Galactosemia type III results from inheriting mutated GALE genes, which causes a partial loss of activity in the GALE enzyme. Three types of epimerase deficiency exist: generalized, intermediate, and peripheral. Generalized epimerase deficiency is like classic galactosemia in terms of severity. As with galactosemia type II, many newborn screening tests do not screen for epimerase deficiency.

There's **a study that follows galactosemia babies** and infants to see if speech delays can be prevented.

Learn more about **Babble Bootcamp** and enroll today!

Who's Who on Your Child's Healthcare Team

A highly specialized multi-disciplinary team can help evaluate and treat your child following a galactosemia diagnosis, and work with you to ensure quality lifelong care. Team members may include the following:

Pediatrician: Your baby's first doctor plays an integral role in his/her care. A pediatrician is trained to diagnose, treat, and provide ongoing care for a variety of childhood illnesses, from mild to severe.

Metabolic Geneticist: These experts specialize in metabolic disorders. They help confirm a diagnosis of galactosemia when your newborn's screening test is abnormal.

Dietician: A dietician can provide guidance on your child's galactosemia-friendly diet, including identifying appropriate substitutes for dairy-based formula or breast milk.

Neonatologist: Some babies will be in the neonatal intensive care unit (NICU). The primary provider is the neonatologist who specializes in the care of newborns with serious illnesses.

Ophthalmologist: It is recommended that an ophthalmologist check for cataracts, which are caused by build-up of toxic galactitol, shortly after your baby's birth. An ophthalmologist is a medical and surgical specialist who deals specifically with the structure, function, diseases, and treatment of the eye, and will monitor your child as she/he grows.

Neurologist: A neurologist is a medical doctor with specialized training in diagnosing, treating, and managing disorders of the brain and nervous system.

Endocrinologist: An endocrinologist is a doctor who specializes in the diagnosis and treatment of hormone-related diseases and conditions.

Social Worker: Social workers support patients and their families through difficult times post diagnosis, with the goal of improving outcomes. They maintain professional relationships and act as guides and advocates for families.

Click here to find a genetic counselor or other specialists in your area.

Fast Fact

The Galactosemia Foundation is led by volunteers! We are always looking for new families to join our team.

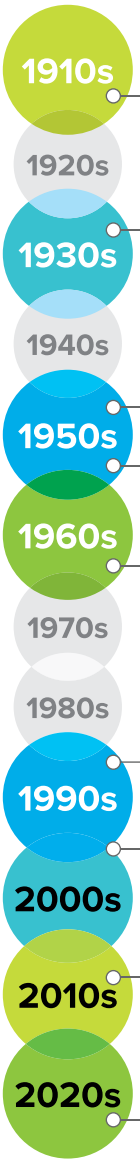
> To learn more, please complete this form.

Did You Know?

The Galactosemia Foundation's **Outreach Team is dedicated to supporting newly diagnosed families** with education, resources, and support.

> Please reach out to us today at outreach@galactosemia.org

Galactosemia Has Been Known for 100 Years, but Identification of Cases has Improved Greatly in Recent Years



1908 — First Documented Case
- Von Ruess, published "Sugar Excretion in Infancy," a case report on a breast-fed infant who failed to thrive, had enlarged liver and spleen, and "galactosuria"
- Infant ceased to excrete galactose when milk products were removed from diet
- Infant later died because of cirrhosis - is believed related to cognac-laced tea therapy

1917 — "Galactosuria"
- Broadly recognized inherited disorder treated by removal of milk products through diet

1935 — First Major Publication
- Mason & Turner, published "Galactosemia: Report of Case with Studies of Carbohydrates"
- First detailed disease description

1950s — "Leloir Pathway"
- Leloir et al. establish metabolic pathway
- Receives Nobel prize for chemistry in 1970

1956 — Discovery of Mutant Gene
- Uncited

1963 — First Newborn Screening ("NBS")
- Guthrie & Paigen
- 2nd disorder detectable through newborn testing

1990 — Waggoner Study
- Waggoner, Bruist & Donnell
- Finds Chronic outcome unrelated to delayed diagnosis/poor dietary compliance

1990s/2000s — Wider NBS Adoption
- Many jurisdictions add CG to NBS
- Eg Ontario, Canada in 2005
- Results in better detection, improved infant survival

2014 — Revised Diet Guidelines
- Van Calcar & Bernstein revise diet guidelines
- All fruits/vegetables allowed
- Aged cheeses allowed

2020 — Revised Diet Guidelines
- The following were changed from restricted to allowed in moderation:
  - Soy sauce, soy products that are fermented (e.g. miso, —natto, tempeh, tofu)
  - Meat by-products — Offal
- First clinical trial for galactosemia begins

For more details on international clinical guidelines, visit www.ncbi.nlm.nih.gov/pmc/articles/PMC5306419/

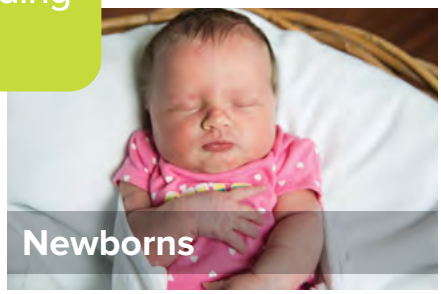
Increased newborn screening is a significant development for the galactosemic community.

Potential Complications

It's important to know that you can do everything right for your child and she/he may still have complications. You can be hypervigilant about ensuring your child adheres to a galactosemia diet and attends all the necessary doctor appointments with your multi-disciplinary team, and still face harsh realities of the disease. Galactosemia is a highly individualized journey; some children have fewer symptoms and complications, and others have more.

Galactosemia is a lifelong condition that presents differently depending on age.

Visit www.galactosemia.org for more information on potential complications by age group and to watch some videos shared by other families who may have experienced what you are going through now.



Additional Resources

Premature Ovarian Failure

Infertility Resources

Understanding Possible Complications Associated with Galactosemia

Even with a restricted diet, our bodies create galactose (referred to as endogenous galactose). Since people with galactosemia cannot break down the galactose, it can lead to complications, including the following:

Cataracts

Between 10-30 percent of newborns diagnosed with classic galactosemia develop cataracts in the first few days or weeks of life. Once your newborn is put on a galactose-restricted diet, cataracts usually clear up on their own, but surgery may be necessary in some cases.

Learning Disabilities

Although no one really knows exactly why, learning disabilities are associated with classic galactosemia. Even children who were diagnosed relatively quickly after birth and adhere to the restricted diet can develop learning disabilities.

Neurological Impairments

Along with well documented speech and language disorders, some galactosemic kids have neurodevelopmental delays.

Primary Ovarian Insufficiency (POI)

Many girls/women who have classic galactosemia experience Primary Ovarian Insufficiency.

Speech Disorders

It is believed that approximately 60 percent of classic galactosemic children have speech problems, ranging from mild to moderate to severe.

Click here for more information on possible complications associated with galactosemia.

Speech Disorder Website

Apraxia-Kids

> A comprehensive information site about childhood apraxia of speech.

Websites

Latest Dietary Guidelines

The Food Allergy Network

Food Allergy Survivors Together (FAST)

Fast Fact

The **Galactosemia Patient Registry** was designed to improve the lives of people living with galactosemia. It is an online system that collects, stores, and retrieves patient data for research studies.

Since galactosemia is so rare, it is important to participate so we can gather the most accurate and up-to-date data in the quest for a cure.

> Learn more and enroll today!

Most complications can be managed but early intervention is key. Find out what early intervention services are available in your state.

> Early Intervention Contact Information by State | CDC

Dietary Guidelines and Resources

Staying up to date on the latest dietary guidelines for galactosemia is critical for the overall care and well-being of your child. Your own clinic/dietician is always your best resource for guidance best suited for your child, but we have compiled additional references below as a starting point.

Since food products change on a regular basis it is imperative that you re-read every label every time you buy it from the grocery store.

Click here for more information about dietary guidelines.

Go Dairy Free

Did You Know?

The Galactosemia Gazette, published online twice a year, is the official newsletter of the Galactosemia Foundation. Newsletter content focuses on current galactosemia research projects; diet, education, and fundraising opportunities; volunteer opportunities and our calendar of events.

Subscribe today!

Top Three Ways to Advocate for Your Child

Raising a child with a rare disease few people recognize or understand can be extremely difficult. Oftentimes you will become an advocate without even realizing it—educating loved ones, friends, and community members about galactosemia to keep your child(ren) safe—and this skill can be readily transferred to advocacy with policymakers, regulators, and the media.

To heighten understanding, research, and funding for galactosemia in the quest for a treatment and, ultimately, a cure, we need to make our voices heard...and that can be as **easy as 1, 2, 3.**

1 Connect with other galactosemia and rare disease families.

By engaging with the Galactosemia Foundation and other rare disease organizations, you can be a part of our collective voice for change. Ten years ago, there were no clinical trials for galactosemia. Now, we have multiple. Help us keep the momentum going!

2 Tell your story.

Your willingness to share what it's like to care for a child with galactosemia is the most effective way to highlight the need for a treatment. Your personal story can poignantly underscore the reasons why people living with galactosemia deserve the same attention, research, and resources as those with more common diseases. Lawmakers, regulators, and reporters need to hear from community members about causes they champion.

3 Be present.

There are fewer families with children with galactosemia, so we need to stick together. Attend the Galactosemia Foundation conference, volunteer for a committee, help a newly diagnosed family along after you hit your stride post diagnosis (believe us, that day will come). It all makes a difference!

You can find the Galactosemia Foundation on **Facebook**, **Twitter**, and **Instagram**.

Get social and follow us today!