

**A PARENT'S GUIDE TO**

# **Understanding Galactosemia**



# A problem with processing

Classic Galactosemia is a condition in which the body has trouble processing a type of sugar called galactose.

Galactose is found in milk, dairy products, and certain other foods. It is also made by the human body, even in people who never eat anything with galactose in it.

Ordinarily, galactose is turned into glucose to give our cells energy. In Galactosemia, this doesn't happen.

Galactosemia is present at birth, caused by a lack of a key enzyme (GALT) needed to process galactose. Enzymes convert molecules in the body.

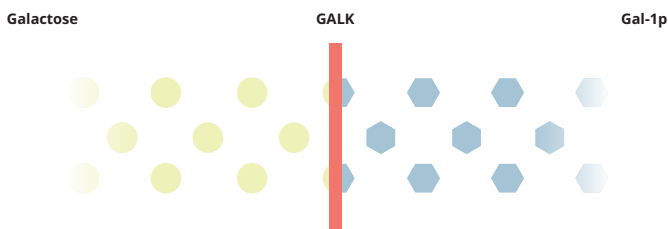
Because galactose can't be processed properly, it is turned into a toxic substance that can damage the body.



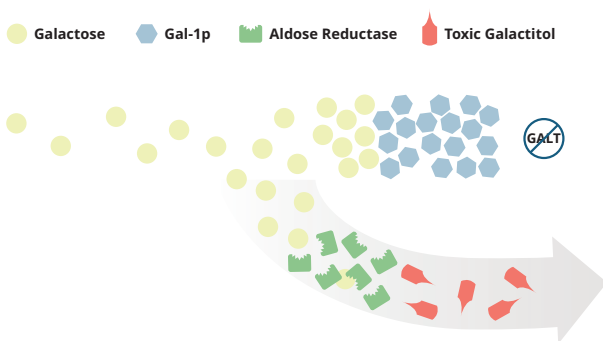
# A tale of two pathways

**A metabolic pathway is where chemical reactions convert molecules in the human body**

**Step 1:** When everything is working the way it should, galactose is converted by Pathway #1. In this pathway, galactose is turned into galactose-1-phosphate, or Gal-1p, by the GALK enzyme.



**Step 2:** Because of the inactive or missing GALT enzyme, Gal-1p can't be processed any further. Gal-1p clogs the pathway, so galactose can't go where it's supposed to.



**Step 3:** Backed-up galactose overflows into Pathway #2, where it shouldn't go. An enzyme that's not supposed to be involved—Aldose Reductase—changes galactose into toxic galactitol.

# The trouble with galactitol

In Galactosemia, galactose is mistakenly turned into a substance called galactitol. We're still learning about galactitol, but what we do know is that it's highly toxic. That 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 problems that people with Galactosemia may experience.



**Cataracts**



**Speech  
issues**



**Cognitive and  
developmental  
delays**



**Motor  
issues**



**Fertility  
issues**



**Tremors**



**Seizures**

# Managing Galactosemia

Most people with Galactosemia follow a special diet to reduce the intake of galactose. Currently, this is the only tool we have to manage the condition.

Because the body makes galactose on its own and this galactose gets turned into toxic galactitol, diet alone isn't enough.

**As scientists learn about the role galactitol plays, this knowledge is changing how we approach Galactosemia.**





[www.galactosemia.com](http://www.galactosemia.com)

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Sources: Berry GT. Classic galactosemia and clinical variant galactosemia. In: Adam MP et al, eds. *GeneReviews*®. University of Washington: Seattle, WA: 993-2020; Berry GT et al. *Lancet*. 1995;346(8982):1073-1074; Coelho A et al. *J Inherit Metab Dis*. 2017;40(3):325-342; Rubio-Gozalbo ME et al. *Orphanet J Rare Dis*. 2019;14(1):86; Welling L et al. *J Inherit Metab Dis*. 2017; 40(2):171-176.