

## Overview of Newborn Screening for Galactosemia Variants – For Parents

### What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions, including **classic galactosemia** and its **variants** (other forms). Babies who screen positive for galactosemia or a galactosemia variant need follow-up tests done to confirm they have one of the types of galactosemia. **Not all babies with a positive newborn screen will have a type of galactosemia.**

### What is galactosemia?

Galactosemia is an **inherited** (passed from parent to child) condition that changes the way a person's body uses a sugar called **galactose**. Galactose is part of another sugar called **lactose** (also called "milk sugar"), which is the main type of sugar found in milk and other milk products.

A person with **classic galactosemia** is missing an **enzyme** (a protein that helps our bodies function) called **galactose-1-phosphate uridyl transferase** (also called **GALT**). When this enzyme does not work properly, galactose cannot be digested (or broken down). Because people with galactosemia cannot break down galactose, it builds up in their blood.

People with **galactosemia variants** have low levels of GALT. The most common galactosemia variant found on newborn screening is called **D/G galactosemia**, where a person has 25 percent GALT activity.

### What causes galactosemia?

Everyone inherits two copies of the GALT gene (one from our fathers and one from our mothers). Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly. In order for a person to have galactosemia, he or she must have two GALT gene changes. People with one GALT gene change are healthy.

### What are the symptoms of galactosemia?

Every child with galactosemia is different. Most babies with galactosemia will look normal at birth. People with galactosemia variants usually have symptoms that are milder (less severe) than classic galactosemia.

Symptoms of classic galactosemia can appear shortly after birth if a baby with galactosemia does not receive treatment. Some of the symptoms of untreated galactosemia include feeding problems, infection, diarrhea, vomiting, liver damage, and cataracts (cloudiness in the eyes), and poor weight gain or growth.

### What is the treatment for galactosemia?

There is no cure for galactosemia. However, there is a treatment that can help with the symptoms. Babies and children with galactosemia should follow a diet that is free of galactose and lactose. Some foods that contain galactose or lactose are:

- Milk and milk products
- Any foods or drugs that contains lactose, casein, caseinate, lactalbumin, curds, whey, or whey solids

People with Duarte or D/G galactosemia may only need to be on this treatment for the first year (12 months) of life.

***If your baby had a positive newborn screen for galactosemia or a galactosemia variant, he/she should start drinking a soy-based formula immediately.***

### What happens next?

Although there is no cure for galactosemia, good medical care makes a difference. Children with galactosemia should see a metabolic geneticist (a doctor who specializes in galactosemia and other related conditions) as well as their pediatrician. Your child's doctor will work with the metabolic geneticist to coordinate any treatment, extra tests, or appointments that your child needs.

### Where are Indiana's metabolic genetics clinics?

Indiana's metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

### Where can I get more information about galactosemia and its variants?

- **STAR-G** - <http://www.newbornscreening.info/Parents/otherdisorders/Galactosemia.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>