

## Galactosemia

(ga-lac-toe-see-me-ah)

### What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called galactosemia. There are other forms of galactosemia that can also cause a positive result on newborn screening. These forms are very rare and are called Galactose Epimerase Deficiency and Galactokinase Deficiency.

**A positive newborn screen does not mean your baby has galactosemia, but it does mean your baby needs more testing to know for sure.**

Your baby's doctor will help arrange for more testing with specialists in disorders like galactosemia.

### What is galactosemia?

Galactosemia affects an enzyme needed to break down a sugar that is part of all milk products, including breast milk and most formulas. Galactosemia is a much more serious problem than lactose intolerance.

A person who has galactosemia doesn't have enough enzyme to break down the sugar known as galactose. If galactose is not broken down, it builds up in the body and causes problems.

Galactosemia is a disorder that is passed on, or inherited, from a child's mother and father. Because galactosemia is a genetic disease, family members are at risk of having galactosemia too, even if no one in the family has had it before.

### What problems can galactosemia cause?

Galactosemia is different for each child. Some children have mild galactosemia with only a few health problems, while other children may have serious complications.

If galactosemia is not treated, a child might develop:

- Serious infection
- Liver failure
- Mental retardation
- Poor growth
- Cataracts (cloudiness) in the eyes

Children and adults treated for galactosemia may still have problems with:

- Speech and language
- Vision (eyesight)
- Growth
- Learning

It is very important to follow the doctor's instructions for testing and treatment.

### What is the treatment for galactosemia?

Galactosemia can be treated. The treatment is life-long. Treatment for children with galactosemia includes:

- Restriction of milk or foods that have milk in them - a dietician will help you set up the best diet for your child

Children with galactosemia should see their regular doctor, a doctor who specializes in galactosemia, and a dietician.

Children with galactosemia can benefit from prompt and careful treatment.

### Resources for parents

**Genetics Home Reference**  
<http://ghr.nlm.nih.gov>

**Parents of Galactosemic Children**  
[www.galactosemia.org](http://www.galactosemia.org)

**MN Children & Youth with Special Health Needs**  
(651) 201-3650 or (800) 728-5420

**MN Newborn Screening Program**  
[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155, Phone (800) 664-7772, Fax (651) 201-5471