

What is Galactosemia?

Galactosemia is a disorder that occurs when the body is unable to break down a type of sugar called galactose that is found in milk and milk products. Instead of being changed into glucose, which the body uses for energy, galactose builds up and can cause damage to the brain, eyes, liver and kidneys.

What causes Galactosemia?

Galactosemia is inherited from both parents. It cannot be “caught” like a cold and cannot be “given” to another child.

How is Galactosemia found?

Shortly after birth, several drops of blood are taken from a baby’s heel. The dried blood sample is sent to the State Department of Health and Senior Service’s Inborn Errors of Metabolism Laboratory, where it is tested for galactosemia and several other conditions. These tests are all part of the State’s Newborn Screening Program. If there is any abnormality, the baby’s doctor and parents are notified.

What does the lab look for?

The lab tests the baby’s blood for the enzyme that should normally be present to break down galactose. The lab also looks to see if the galactose level is elevated.

Does an abnormal screening test mean that my baby has Galactosemia??

Screening tests always need to be confirmed by additional testing and medical evaluation.

IF YOU ARE ASKED TO HAVE YOUR
BABY RE-TESTED, ACT QUICKLY AND
FOLLOW YOUR DOCTOR’S ADVICE

What will my baby’s doctor do?

The doctor will immediately place the baby on a soy formula. Breastfeeding must be stopped until the results of the repeat testing are known. The doctor may refer the baby to a metabolic/genetic specialist. The doctor and specialist will confirm the diagnosis by doing the following:

- Getting a complete medical history and doing a physical examination.
- Looking for symptoms such as vomiting, diarrhea, failure to grow and gain weight yellowing of the skin and eyes
- Getting another blood sample and a urine sample to do special tests that confirms the screening results and determines the diagnosis.

How is Galactosemia treated?

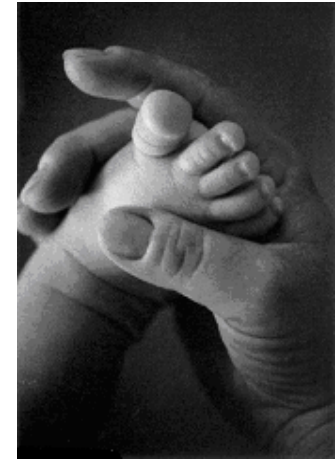
Galactosemia is treated for life with a special diet. Any food that has galactose or lactose cannot be eaten (milk or milk products). Frequent medical check-ups are essential to monitor the baby’s growth and development.

For more information, contact:

The New Jersey Department of Health
and Senior Services
at (609) 292-1582

or

The Inborn Errors of Metabolism
Laboratory at
(609) 292-3090



Galactosemia

Important Information
For Parents



