

Galactosemia

What is the Wisconsin Newborn Screen?

All babies in Wisconsin have a test called the Newborn Screen (NBS) in their first 24 hours of life. A blood sample is taken from baby's heel for this test. This test looks for rare medical conditions that may not be caught or show up in your baby right away.

Newborn screening gets babies the help they need by:

- Finding out they have a condition before they have any medical problems
- Allowing treatment to be started right away
- Preventing medical emergencies

What was my child's NBS result?

Your baby's NBS has screened positive with a high level of a marker called "galactose" and low level of an enzyme called "galactose-1-phosphate uridyl transferase" (GALT) This means your baby may have a condition called galactosemia.

This test is a screening test. We will need more information to find out if he or she has this diagnosis.

What is galactosemia and why is it important?

Galactosemia affects how the body breaks down a sugar called galactose that is found in human milk, many baby formulas, and other dairy products. Babies with galactosemia can become sick very quickly when they get galactose in their diet. Symptoms include difficulty eating, staying awake, or responding to you, not gaining weight, and jaundice (yellowing of the eyes and skin).

What are the next steps?

Your baby's doctor will give you instructions about what your baby should be eating and may order that he or she switch to soy formula (which does not have galactose). You should continue the soy formula until your doctor says it is safe to stop or until you are seen in the Genetics clinic and a dietitian helps you come up with a new feeding plan.

Tests and Doctor's Visits

This will help us know if your baby has galactosemia or not. Your baby's health care provider will order:

- An urgent visit to the primary care clinic and/or emergency department
- Blood and urine (pee) tests
- Referral to the Genetics Clinic is possible

What if I have questions or concerns?

Contact your primary care provider right away: If your baby is not eating well, gets sick, or you have questions or concerns about how he or she is doing.

Call the Children's Wisconsin Genetics Center: If you have questions about Newborn Screen, galactosemia, or your baby's test results.

- Call 414-266-3347, option 3
- Leave a message and a nurse or genetic counselor from the Newborn Screen program will call back.
- Office is open Monday-Friday, 8 AM-4PM

Scan the QR codes below for links to more information

Medline Plus
Genetics:



NewbornScreening.Info:

