NORTH CAROLINA NEWBORN SCREENING PROGRAM

Galactosemia Fact Sheet for Parents

All babies born in North Carolina are screened at birth for certain medical conditions that can be treated to prevent serious illness. This is called newborn screening. The newborn screening result for your newborn showed that your baby might have galactosemia (guh-lak-tuh-see-mee-uh). Your baby will be referred to a specialist for more testing.

There may not be signs of galactosemia at birth.

What is Classic Galactosemia?

Classic galactosemia is a rare inherited disorder that affects your child's ability to digest milk and dairy. Individuals with classic galactosemia have a deficiency (not enough) of the galactose-1-phosphate uridyltransferase (GALT) enzyme. This means that they are unable to break down and use galactose, a sugar found in milk and dairy products. As a result, the child cannot breakdown and use breast milk or cow's milk. If treatment is not started early, life threatening complications can occur.

What are the symptoms of Classic Galactosemia?

Individuals with classic galactosemia may have a range of symptoms, or signs of the disease. Symptoms typically appear within the first week of life and can include:

- Poor feeding
- Vomiting
- Yellowish skin and whites of eyes (jaundice)
- Lack of energy (lethargy)
- Bleeding
- Bulging on the top of the head (bulging fontanel)
- Cataracts (haziness of the lens of the eye)

What is Duarte Galactosemia?

Duarte galactosemia is a mild form of galactosemia that is unlikely to show symptoms and does not require treatment. This condition is sometimes identified as part of newborn screening for classic galactosemia.

What happens next?

Your baby's pediatrician may refer your baby to a specialist for follow-up testing depending on your baby's specific newborn screen result. In the meantime, your pediatrician may ask you to switch to a soy-based formula.

How is Galactosemia treated?

Treatment for babies suspected to have **classic galactosemia** include:

- **Avoid foods containing galactose:** This includes breast milk and dairy. Use soy-based baby formula instead. A change in mom's diet does not affect the level of galactose in breast milk.
- **If you are concerned about any symptoms in your baby, contact your baby's pediatrician immediately. If your newborn has symptoms of galactosemia, he/she may need to go to the hospital.**

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Baby's First Test: https://www.babysfirsttest.org/newborn-screening/conditions/classic-galactosemia

Genetic and Rare Diseases Information Center: https://rarediseases.info.nih.gov/diseases/13639/classic-galactosemia

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