NORTH CAROLINA NEWBORN SCREENING PROGRAM

Galactosemia Fact Sheet for Providers

What is Classic Galactosemia?

Classic galactosemia is characterized by an inability or reduced ability to metabolize galactose, a sugar found in lactose, due to an inherited deficiency of the galactose-1-phosphate uridyltransferase (GALT) enzyme. This leads to elevated levels of galactose and galactose-1-phosphate (gal-1-P) in the blood. Due to the presence of high levels of galactose in both breast milk and cow milk formulas, the condition can be life threatening if treatment is not started early.

What are the signs and symptoms of Classic Galactosemia?

Infants with classic galactosemia may show no symptoms at birth. If they are symptomatic or ill, they should be transferred to a hospital for treatment. Possible symptoms include:

- Jaundice
- Poor feeding
- Vomiting
- Lethargy
- Bulging fontanel
- Bleeding
- Cataracts
- Signs of sepsis

Duarte Variant

The Duarte variant of galactosemia is generally regarded as a benign condition without symptoms. In this variant there is only a slight reduction of normal GALT activity, not the complete or almost complete deficiency found in classic galactosemia. This results in some difficulty metabolizing galactose in early childhood. Although these infants typically do not need to be switched to a soy formula, we recommend meeting with the geneticist to discuss this benign disorder.

How is classic galactosemia identified and diagnosed?

All infants with an abnormal galactosemia newborn screening result concerning for classic galactosemia need:

- A clinical examination with a genetics team
- Confirmation with enzyme assay and measurement of gal-1-P
- Molecular genetic testing to confirm the diagnosis.

How do I handle an abnormal screen for galactosemia?

Early diagnosis is very important for classic galactosemia, as the condition can be fatal without treatment. In the case of an abnormal screening, IMMEDIATELY take the following steps:

- Follow the instructions faxed by the North Carolina Newborn Screening Coordinator regarding follow up testing, referrals and treatment. Reach out to the NBS Coordinator if you have additional questions.
- If the baby is displaying any of the symptoms listed above, consult with a metabolic or genetic specialist and arrange to transfer them to a hospital for further treatment.

How is Galactosemia treated?

- Treatment for classic galactosemia consists of avoiding breast milk, dairy products, and other food containing galactose. These can be substituted with soy-based formulas for infants. No change to the maternal diet will alter the galactose content in the breast milk.
    - If the baby is symptomatic, emergency medical measures will need to be taken.
- Usually no treatment is required for the Duarte variant.

Where do I go for more information?

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

UNC Health Information and Referrals: https://www.uncchildrens.org/uncmc/unc-childrens/care-treatment/genetic-disorders/referrals/

ACMG Classic Galactosemia ACT Sheet: www.acmg.net/PDFLibrary/Classical-Galactosemia-ACT-Sheet.pdf

MedlinePlus: https://medlineplus.gov/genetics/condition/galactosemia/

Where do I send parents for 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

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

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