NORTH CAROLINA NEWBORN SCREENING PROGRAM

GALE, GALK and GALM Deficiency Fact Sheet for Providers

What are GALE, GALK and GALM Deficiency?

This conditions group of genetic disorders is characterized by an inability to metabolize galactose, a sugar found in lactose. Unlike classic galactosemia, this is not caused by a deficiency of the galactose-1-phosphate uridyl transferase (GALT) enzyme, but a lack of either the UDP-galactose-4 epimerase (GALE), galactokinase (GALK), or galactose mutarotase (GALM) enzymes. The exact symptoms and severity of the disorder vary based on the specific enzyme affected.

Due to the presence of high levels of galactose in both breast milk and cow milk formulas, these conditions can lead to long-term health problems if not treated early.

What are the symptoms of these conditions?

Infants with these conditions are usually asymptomatic in the first weeks of life.

GALE deficiency: If untreated, symptoms can include:

- Liver dysfunction leading to hepatomegaly and jaundice
- Poor feeding and emesis
- Cataracts

GALK and GALM deficiency: If untreated, symptoms can include:

- Cataracts

How are these conditions identified and diagnosed?

All infants with an abnormal newborn screening result concerning for these conditions need a clinical examination and confirmatory testing. If classic galactosemia (GALT deficiency) has been ruled out based on NBS GALT level, but total galactose levels are elevated, infants may require evaluation for GALE, GALK or GALM deficiency. Biochemical confirmation includes GALK and GALE enzyme activity assays, as well as galactose-1-phosphate levels in erythrocytes, and galactitol in plasma or urine. Molecular genetic testing should be performed in all cases of suspected GALE, GALK or GALM deficiency.

How do I handle an abnormal screen for GALE, GALK or GALM deficiency?

In the case of an abnormal screen, take the following steps:

- Discontinue breast feeding and cow milk formulas until GALE deficiency can be ruled out.
- Check infant for symptoms

If you are notified that one of your patients received a positive newborn screening result concerning for GALE, GALK or GALM deficiency, make an appointment at the genetics center of your choice. Refer to the fax received from the NBS follow-up for a list of centers.

How are GALE, GALK or GALM deficiency treated?

Place infant on a galactose-free (or soy-based) formula such as Prosobee, Isomil and Nutramigen.

GALE deficiency: Patients with GALE deficiency are treated with a galactose free diet.

GALK and GALM deficiency: Patients with GALK and GALM deficiency are treated with a galactose-reduced diet.

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/

National Library of Medicine:
https://www.ncbi.nlm.nih.gov/books/NBK51671/

Where do I send parents for information?

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

Baby's First Test: GALE deficiency
https://www.babysfirsttest.org/newborn-screening/conditions/galactoepimerase-deficiency

Baby's First Test: GALK deficiency
https://www.babysfirsttest.org/newborn-screening/conditions/galactokinase-deficiency

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