

GALE, GALM and GALK Deficiency Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain diseases or other health problems that can be treated if caught early. The newborn screening result showed that your baby might have a condition affecting the breakdown of galactose, a sugar found in foods. Your baby will be referred to a specialist for more testing to see if they have one of these conditions.

What are GALE, GALK and GALM deficiency?

Galactose is a sugar found in milk and dairy products. Galactose is broken down in the body by enzymes. When these enzymes don't work properly, a baby can have conditions called GALE deficiency, GALK deficiency, and GALM deficiency. The specific diagnosis depends on which enzyme is not working properly. These conditions usually do not have lasting effects, but may require treatment to prevent symptoms. There often are no signs of these conditions at birth.

What are the symptoms of these conditions?

GALE deficiency: If untreated, symptoms can include:

- Liver dysfunction causing liver enlargement and jaundice (yellowing of the skin and whites of eyes)
- Poor feeding and vomiting
- Cataracts

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

- Cataracts

What happens next?

Your baby's pediatrician will refer your baby to a specialist for follow-up testing. These tests will check your baby's blood to find out if your baby has any of these conditions. Your baby may be put on a diet of soy-based formula, not breast milk or cow milk formula, while you wait for the results of these tests. With an early diagnosis and careful diet, damage can be prevented, and your child can grow to live a healthy life. Doctors, dieticians and genetic counselors work closely with families to manage the condition.

How are these conditions treated?

GALE deficiency: People with GALE deficiency are treated with a galactose free diet. This means avoiding most dairy and other high lactose foods. Infants are switched to a formula that does not contain galactose.

GALK and GALM deficiency: People with GALK and GALM deficiency are treated with a galactose-reduced diet. This means limiting most dairy and other high lactose foods. Infants are switched to a formula that does not contain galactose.

Where do I go for more 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/galactosepimerase-deficiency>



Baby's First Test: GALK deficiency

<https://www.babysfirsttest.org/newborn-screening/conditions/galactokinase-deficiency>

