

Positive Result:

Blood Spot Screen Result Notification



High Levels of Total Galactose (TGAL) with Normal Levels of Galactose-1-Phosphate Uridyltransferase (GALT)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of total galactose (TGAL), but normal levels of galactose-1-phosphate uridyltransferase (GALT).

What does this mean?

High levels of TGAL with normal levels of GALT can indicate that your child may have galactokinase (GALK) or galactose epimerase (GALE) deficiency. A positive result does not mean your baby has GALK or GALE deficiency, but more testing is needed to know for sure if one of these enzymes is abnormal.

What happens next?

Your baby's doctor will help arrange for more testing and schedule an appointment with a metabolic specialist. If your baby has GALK or GALE deficiency, this testing will help figure out which one they have since newborn screening cannot tell the difference.

The specialist will likely want to switch your baby to a soy-based formula since breastmilk and milk-based formulas can cause health problems in children who have GALK or GALE deficiency.

What is GALK and GALE deficiency?

GALK and GALE deficiency are rare conditions present in a baby at birth where the body is not able to use a sugar called galactose. Galactose comes from food, including all breastmilk, all dairy products, and many baby formulas. People with GALK or GALE deficiency have too much galactose in their blood. Too much galactose in the blood can cause health problems.

What health problems can it cause?

If left untreated, children with GALK deficiency can develop cataracts.

There are three forms of GALE deficiency. Most individuals with GALE deficiency have the mild form that usually causes no health problems.

What treatment options are available?

Although GALK deficiency cannot be cured, it can be treated. Many children with GALK deficiency need to avoid all milk products.

Treatment is not always needed for children with GALE deficiency. If treatment was recommended, it would involve avoiding milk products.

Children with GALK or GALE deficiency should see their regular doctor and a metabolic specialist.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org