

Positive Result:

Blood Spot Screen Result Notification



Absent/Reduced 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 no or low levels of an enzyme called galactose-1-phosphate uridyltransferase (GALT).

What does this mean?

Absent or low levels of GALT can indicate that your child may have galactosemia. A positive result does not mean your baby has galactosemia, but more testing is needed to know for sure.

What happens next?

Your baby's doctor will help arrange for more testing with specialists familiar with galactosemia. The specialists will want to see your child as soon as possible to get labs drawn.

The specialists 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 classic galactosemia.

What is classic galactosemia?

Classic galactosemia is a condition present in a baby at birth where the body is not able to use a milk sugar called galactose. Galactose comes from food, including all breast milk, all dairy products, and many baby formulas. The term 'galactosemia' literally means too much galactose in the blood.

What health problems can it cause?

Classic galactosemia is a lifelong condition. If left untreated, it can cause:

- Poor feeding and weight gain
- Lack of energy
- Jaundice (yellow skin or whites of the eyes)
- Liver damage
- Brain damage
- Overwhelming bacterial infections (sepsis) that can be life-threatening

Children with classic galactosemia can benefit from prompt and careful treatment.

What treatment options are available?

Although classic galactosemia cannot be cured, it can be treated. The most common treatment is a change in diet to avoid all milk products. If treated before symptoms develop, children can have healthy growth and development.

Children with classic galactosemia 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