Family action sheet for galactosemia

If a newborn blood screening result shows that your baby could have a serious condition called galactosemia, this result does not mean that your baby has this condition, but more tests are needed.

All babies born in California have a routine blood screen shortly after birth. The goal of this newborn screening is to find those at risk for serious medical conditions. Babies can look healthy at birth and still have one of these conditions. Babies with these conditions benefit from early diagnosis and treatment.

What is galactosemia?

Babies with this disorder lack an enzyme needed to break down the sugar in milk. Without treatment, sugar called galactose can build up in the body and cause damage. There are different forms of galactosemia based on gene changes that cause the condition, called variants. Genes are code in our cells passed down from parents to babies. They make us who we are.

Babies with classic galactosemia have symptoms soon after birth if they receive too much galactose (either from breast milk or dairy-based formula). If left untreated, infants with galactosemia can develop very serious symptoms.

Is there treatment for galactosemia?

Yes, there are treatments for galactosemia. If found early, the treatments are more successful.

Your medical provider or a galactosemia specialist will tell you if your baby has it and will discuss a treatment plan. It might mean that your child will need a low-galactose diet.

Newborn screening takes blood from a newborn's heel to screen for medical conditions

Next steps

1. **Your baby's medical provider will order more testing as soon as possible**
   These tests will let you know if your baby has galactosemia. While waiting for additional test results, your medical provider may recommend stopping dairy-based formula products and breast feeding.
2. **Work closely with your baby's medical provider and follow recommendations**
   Your infant's medical provider may refer your infant to a doctor with more experience in galactosemia. It is important that you keep all appointments and start treatment if instructed.

Where can I get more information?

Your infant's medical provider or galactosemia doctor is the best person to teach you about galactosemia. Find more information at these websites:

- **Baby's First Test** (https://www.babysfirsttest.org). Type "galactosemia" into the "Find a Condition" Box. Follow the link. For Spanish, click on the top right black box marked "en Español"
- **Genetics Home Reference** (https://ghr.nlm.nih.gov/condition/galactosemia)

California Department of PublicHealth

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)

CALIFORNIA GDSP Genetic Disease Screening Program