

PARENT INFORMATION for Inconclusive newborn screening results for GALACTOSEMIA

When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually cannot be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were "inconclusive" for a disorder called Galactosemia (Guh-LAK-toe-SEE-me-ah). It is important to remember this is only a screening test. A new blood sample will be collected from your baby and tested to help find out if your baby has galactosemia or not.

What does an inconclusive screen mean?

A small percent of all babies screened will be identified as "inconclusive" on screening. Most are later found not to have the disorder. An inconclusive screen means the test could not rule out Galactosemia. A repeat screening test should be done to help determine if your child has this disorder or not. Keep in mind that the repeat test could show that your baby does not have galactosemia.

What is the next step?

Be sure to follow your baby's doctor's instructions for getting a new blood sample collected for further testing. Galactosemia is a rare metabolic disease, so your baby's doctor might consult with or refer you to a specialist.

What exactly is Galactosemia?

Galactosemia is a disease in which galactose cannot be broken down in the body. Galactose is a simple sugar found in breast milk, many formulas and milk products. Children with galactosemia

cannot break down the galactose, so it remains at high levels in their bodies. The effects of these high levels can harm a child's eyes, liver, and brain. About 1 in 4 infants with untreated galactosemia develop serious infections in the first or second week of life. If necessary, diet changes within the first few days of life can minimize this damage.

Finding out early if your baby has galactosemia, allows you and your baby's doctor to take steps to prevent most of these effects.

What should I do?

Three things to do immediately:

1) Take your baby to have the new blood sample collected and tested. 2) Report to your baby's doctor immediately, if your baby should show any signs of illness such as fever, vomiting, or diarrhea.

If your baby does have galactosemia, there are very good treatments available. The pediatric metabolic specialists and nutritionists will be able to help you and your baby's doctor manage this through checking the blood levels and adjusting the diet.

You have probably already done this, but now is also a good time to be sure your newborn is covered by your health insurance provider. Most health insurance covers all or part of the medical tests needed for treating galactosemia.

For more information talk with your baby's doctor or you can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on Pediatric Metabolic Specialists available in Nebraska.

