PARENT INFORMATION for POSITIVE 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 positive 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 a new test will be done to help find out if your baby has galactosemia or not.

What does a positive screen mean?

A positive test means that your baby needs to have a new blood test to check for galactosemia. A small percent of all babies screened will be identified as "positive" on screening. Some are later found not to have the disorder. This is just a screening test and further testing needs to be done to determine if your child has this disorder or not. Keep in mind that the new 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 for further testing. Because newborns with Galactosemia can become sick very quickly from too much galactose, they should be switched to a powder-based soy formula as soon as the new blood sample is drawn. Galactosemia is a rare 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. Placing the child on a powder based soy formula 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) Begin feeding your baby a powder-based soy formula after the blood is drawn. 3) Report to your baby's doctor immediately, if your baby should show any signs of illness such as fever, vomiting, or diarrhea.

If it turns out your baby does have galactosemia, there are very good treatments available. The pediatric metabolic specialists and nutritionists will be able to help you 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.

Nebraska Department of Health and Human Services

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