Arkansas Department of Health
Newborn Screening Result: Decreased GALT activity
Interpretation Sheet for Parents
Infant may have Variant Galactosemia

You have just heard that your infant may have Galactosemia. Please understand that the newborn screening is just that – a screening test. Further testing is required to confirm or rule out the diagnosis.

Overview

Galactosemia which means "galactose in the blood," is a rare inherited condition in which the body is unable to digest a type of sugar called galactose. Galactose is found in milk and many other types of food, but it cannot be used properly in children who lack an enzyme needed to break down galactose. Signs and symptoms of variant galactosemia depend on the level of activity of the enzyme that breaks down galactose. The name of the enzyme is galactose-1-phosphate uridyl transferase. It is also known as GALT or Gal-1-PUT.

What is Variant Galactosemia?

Variant Galactosemia is caused by decreased activity of a liver enzyme (GALT/Gal-1-PUT.) required to use galactose. Because galactose cannot be broken down, it builds up in the cells and may cause problems.

Does a positive newborn screening result mean that a baby has Galactosemia? Not always. Some babies who are screened will be identified as "positive" on screening, but later found to not have the disorder. Further testing needs to be done in order to determine if the baby has the disorder, or to rule it out as a "false positive."

How is Galactosemia diagnosed?

After receiving a positive newborn screen, the most important thing parents can do is be sure that their baby goes in for a new specimen to be collected and tested as soon as possible. This sample will be tested for galactosemia using a more precise test.

Why is newborn screening done for Galactosemia?

Newborn screening for Galactosemia offers early detection so that treatment can begin earlier. Early detection of Galactosemia can help prevent many of the complications that may arise if the condition is severe.

What are the signs and symptoms of Variant Galactosemia?

Signs and symptoms of variant galactosemia depend on the level of activity of the GALT enzyme that breaks down galactose.

- If GALT activity is low but adequate, the infant will probably not show any symptoms. The baby can feed on breast milk without any problems.
- If GALT activity is too low, symptoms can include difficulty feeding, lack of energy or irritability. The baby should switch to soy formula.

What should I feed my baby?

Please read the following carefully.

Your baby's doctor might recommend that your infant can continue on breast milk or needs to switch to a soy formula. The recommendations might change depending on the results of diagnostic testing.

- If your infant is on breast milk and is feeding well, and gaining weight, then your baby's doctor will order a urine test. If the urine test is normal, you can continue to breastfeed the baby. If the urine test shows an abnormality, the baby will have to be bottle fed soy formula.

- If your infant is bottle fed, your baby's doctor will recommend that the baby switch to a soy formula.

- If your infant is breast fed and is not feeding well or has vomiting, the baby's doctor may recommend that you bottle feed the baby soy formula.

How long will I have to restrict milk from the baby's diet?

Until your baby is about 1 year old. At that time, your baby will have a blood and urine test to determine how well he/she is using galactose. Almost always your child will be able to have milk and milk products after 1 year of age.

Where can I get additional information?

http://www.arkansas.gov/newborn_screening/index.html