Arkansas Department of Health
Newborn Screening Result: Very Low GALT activity
Interpretation Sheet for Parents
Infant may have Classical Galactosemia

You have just heard that your infant may have Galactosemia. Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will have a diagnosis of galactosemia. Further testing is required to confirm or rule out the diagnosis.

What is Galactosemia?
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. The name of the enzyme is galactose-1-phosphate uridyl transferase. It is also known as GALT, or Gal-1-PUT. If GALT is absent or low, galactose cannot be broken down. It then builds up in the cells becoming toxic. In response, the body is unable to produce energy. This causes the signs of GALT, which include poor feeding, lack of energy, irritability, and convulsions.

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 should the disorder go untreated during a child's infancy. Specialists may be able to improve the quality of life for individuals with Galactosemia.

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 common is severe Galactosemia?
Galactosemia affects one in every 60,000 babies.

What are the symptoms of Galactosemia?
Galactosemia will usually cause no symptoms at birth, but jaundice (yellowing of the skin), diarrhea, and vomiting will quickly develop as the baby fails to gain weight. If you see any of these signs, report them to your baby's doctor immediately. With treatment, normal development is very likely. Without treatment, infections can become common, and the build-up of galactose will cause liver disease, cataracts, and mental retardation. Other symptoms include: poor feeding, lack of energy, irritability, and convulsions.

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 more precise tests.

If the baby does have Galactosemia, there are treatments available. A pediatric specialist known as a metabolic geneticist will help confirm a diagnosis.

How is Galactosemia treated?
The baby's primary doctor will work with a metabolic doctor and a dietician to treat the condition. Galactosemia is treated at first by changing the baby to soy-based formula that is free of galactose. To prevent mental retardation, treatment must begin soon after diagnosis. A person with a confirmed diagnosis of Galactosemia should avoid galactose for life.

Where can I get additional information?
http://www.arkansas.gov/newborn_screening/index.html