Arkansas Department of Health
Newborn Screening Result: Elevated Succinylacetone
Infant may have Tyrosinemia type I
Information Sheet for Parents

You have just heard that your infant may have Tyrosinemia. Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will be confirmed to have the diagnosis. The below information is meant to keep you informed while further testing is done to evaluate the newborn screening result.

What is Tyrosinemia?

Tyrosine is one of the twenty amino acids that our proteins are made of. There are several types of tyrosinemia. Babies with tyrosinemia are unable to use tyrosine. If not treated, tyrosinemia type I causes poor weight gain, vomiting, swelling of legs, and liver/kidney problems. A baby with tyrosinemia may require treatment with a special formula, medication, or even liver transplantation.

Why is newborn screening done for Tyrosinemia?

Newborn screening for tyrosinemia offers earlier detection so that treatment can begin earlier. Early detection of tyrosinemia is key to preventing many of the complications that may arise should the disorder go untreated during a child's infancy. Specialists will be able to improve the quality of life for people with tyrosinemia.

Does a positive newborn screening result mean that a baby has tyrosinemia?
Not always. Some babies who are screened "positive" are later found not to have the disorder after further testing. This is because the screening test is not a diagnostic test. A screening test is designed to identify all infants with the disorder, but sometimes picks up infants who don't have the disorder. Therefore 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 tyrosinemia?
About one in every 100,000 babies in the United States is born with this condition. In people of French Canadian ancestry, tyrosinemia occurs in about 1 out of 12,500 babies.

What are the signs and symptoms of tyrosinemia?
Some babies will appear normal at birth, depending upon the type of Tyrosinemia. Babies will usually show the condition within the first few months of life. Some of the first symptoms may be: diarrhea, vomiting, poor weight gain, extreme sleepiness, irritability, and "cabbage-like" odor to the skin or urine. Liver problems can commonly include: enlarged liver, yellowing of the skin, easy bruising, and swelling of the legs or abdomen. Kidney problems may lead to rickets (a bone thinning condition), and delays in walking.

How is tyrosinemia diagnosed?
After receiving a positive newborn screen, the most important thing parents can do is be sure their baby goes in for a new specimen to be collected and tested as soon as possible. If the baby does have tyrosinemia, there are treatments available. A pediatric metabolic doctor and geneticist will help confirm a diagnosis.

How is tyrosinemia treated?
The baby's doctor will work with a metabolic doctor and dietician to care for the child. Lifelong treatment is usually needed in order to prevent liver and kidney problems. Treatment involves medication and a diet low in tyrosine and another amino acid called phenylalanine. This diet consists of a special medical formula and carefully chosen foods. The treatment is started as soon as the child is diagnosed with the condition.

Where can I get additional information?
http://www.arnewbornscreening.com