

Arkansas Department of Health
Newborn Screening Result: Infant may have Fatty Acid Oxidation Disorder
Information sheet for Parents

You have just heard that your infant may have a fatty acid oxidation disorder. 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.

Overview

Our bodies get energy from the food we eat. If we go for more than a few hours without food or during periods of illness, our body uses fat for energy. We have enzymes that break down some of the fats in food and also the fat that is stored in our body. Babies with fatty acid oxidation disorders are missing one of these enzymes or their enzyme does not work correctly. They will have problems breaking down their stored fat and turning it into energy so they cannot fast (go without eating) for long periods of time. There are different types of fatty acid oxidation disorders depending on which enzyme is missing.

Why is newborn screening done for fatty acid oxidation disorders?

If a fatty acid oxidation disorder is not treated, it can cause serious health problems. Newborn screening allows babies to be detected early so that doctors can prevent these medical problems.

Does a positive result of newborn screening mean that a baby has fatty acid oxidation disorder?

Not always. Some babies who are screened “positive” are 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.”

What are the signs and symptoms of a fatty acid oxidation disorder?

A lot of babies don’t have any symptoms at birth. Symptoms usually begin shortly after birth up until 2 years of age, but may not occur for many years. The symptoms can be very mild in some infants, and serious in others. Fatty acid oxidation disorders usually cause episodes of low blood sugar, which can happen when the baby hasn’t eaten for more than a few hours. Symptoms of low blood sugar include: sleepiness, weakness, poor weight gain, vomiting, behavior changes including irritability, and liver and heart problems

It is therefore important that you feed your baby every 3 hours while waiting for results of the diagnostic test.

If hypoglycemia is not treated, more serious problems can occur such as breathing problems, seizures, brain damage, and coma which could lead to death.

How are fatty acid oxidation disorders 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. The test results should be available within 3 days.

How are fatty acid oxidation disorders treated?

Treatment is usually very effective. The type of treatment will depend on the child. Babies need to eat every few hours, including throughout the night. As the baby gets older, he/she may be able to go longer without eating. When children are ill, they may not feel hungry but is important that they still eat and drink fluids so they don’t develop hypoglycemia. Your child’s metabolic doctor may recommend a diet that is low in fat and high in carbohydrates. Supplements may also be prescribed.

Where can I get additional information?

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