

Arkansas Department of Health
Newborn Screening Result: Elevated C5-OH Acylcarnitine
Interpretation Sheet for Parents
Infant may have Organic Acid Disorder

You have just heard that your infant may have an Organic acidemia. 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 an Organic Acid disorder?

Children with organic acid disorders do not have enough of a specific enzyme that helps break down certain proteins. This leads to an abnormal buildup of particular acids called organic acids. Abnormal levels of organic acids in the blood (organic acidemia), urine (organic aciduria), and tissues can be toxic and cause serious health problems. Most cases of organic acid disorders show symptoms within the first two weeks after birth.

Why is newborn screening done for organic acid disorders?

Newborn screening for organic acid disorders offers early detection so that treatment can begin earlier. Early detection can help prevent many of the complications that may arise should the disorder go untreated during infancy.

Does a positive newborn screening result mean that a baby has an organic acid 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 organic acid disorders?

Symptoms often appear in the first few weeks of life. Symptoms can include a lack of appetite, vomiting, seizures, lack of energy, muscle irritability, and hypothermia. Illness can be triggered by infections or by fasting. If left untreated, organic acid disorders may cause serious medical problems including brain damage, coma and even death.

How are organic acid 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. If the baby does have an organic acid disorder, there are treatments available. A pediatric metabolic doctor and geneticist will help confirm a diagnosis.

How are organic acid disorders treated?

The baby’s primary doctor will work with a metabolic doctor and a dietician to treat the condition. Organic acid disorders are treated with low-protein diet, medical foods, medical formula and avoiding fasting. Sometimes, other protein supplements may need to be added to the baby’s diet. Any changes in diet should be made under the guidance of a dietician. Individuals must follow the special diet for the rest of their lives.

Where can I get additional information?

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

