

**Arkansas Department of Health
Newborn Screening Result: Elevated C5 marker
Infant may have Isovaleric Acidemia
Information Sheet for Parents**

You have just heard that your infant may have Isovaleric Acidemia (IVA). 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 IVA?

Amino acids are the building blocks for proteins found in foods. Isovaleric acidemia (IVA) is a rare inherited disorder in which the body is unable to process one amino acid (leucine) correctly. Children with IVA do not have enough of a specific enzyme that helps break down leucine. It is an organic acid disorder, meaning it 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 IVA show symptoms within the first two weeks after birth.

Why is newborn screening done for IVA?

Newborn screening for IVA offers early detection so that treatment can begin earlier. Early detection of IVA can help prevent many of the complications that may arise should the disorder go untreated during a child's infancy.

Does a positive newborn screening result mean that a baby has IVA?

Probably, though 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 IVA?

IVA affects one in every 100,000 babies.

What are the signs and symptoms of IVA?

Every child is different and some of these symptoms may not apply to your child specifically. 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 low body temperature. Illness can be triggered by fasting, infections or by consuming high-protein foods. A foul "sweaty feet" odor is also very common. If left untreated, IVA may cause serious medical problems including brain damage, coma and even death.

How is IVA 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 IVA, there are treatments available. A pediatric metabolic doctor (specialists in body chemistry) will help confirm a diagnosis.

How is IVA treated?

The baby's primary doctor will work with a metabolic doctor and a dietician to treat the condition. IVA is treated at first by changing the baby to formula that is low in leucine. To prevent brain damage, treatment must begin soon after diagnosis. Any changes in diet should be made under the guidance of a dietician. Supplemental medication may also be used. 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