Arkansas Department of Health
Newborn Screening Result: Infant may have Glutaric Aciduria
Condition description sheet for Parents

You have just heard that your infant may have Glutaric Aciduria Type I (GA-1). 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 glutaric aciduria type I?
Glutaric aciduria type I (GA-1) is a rare inherited disorder in which the body is unable to break down some proteins correctly. Children with GA-1 do not have enough of a specific enzyme that helps break down certain proteins. Two amino acids found in proteins – lysine and tryptophan – are incompletely broken down causing a build-up of glutaric acid, which in turn causes health problems.

Why is newborn screening done for GA-1?
Newborn screening is done for GA-1 so that babies with this condition can be diagnosed quickly. This allows treatment to begin before any major health problems occur.

Does a positive result of newborn screening mean that a baby has GA-1?
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."

How common is GA-1?
About 1 in every 40,000 babies will have GA-1. It is more common in the Amish community.

What are the signs and symptoms of GA-1?
Most babies with GA-1 are healthy at birth. Symptoms usually begin between 2 months and 4 years of age though some infants have no symptoms. GA-1 can cause periods of severe illness, called metabolic crises. "Metabolic" refers to body chemistry. Metabolic crises are often caused if the baby goes for long periods without food, or has a fever or illness. Early symptoms of a metabolic crisis include: abnormal movement, vomiting, low muscle tone and muscle weakness. Intravenous glucose during illness is the most important treatment. If treatment is started early, most metabolic crises and health problems can be prevented. Even with treatment, some children will still have metabolic crises.

How is GA-1 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 using more specific tests. A pediatric metabolic doctor and geneticist will help confirm a diagnosis.

How is GA-1 treated?
If treatment is started early, babies with GA-1 can often grow up to have healthy lives with normal growth and development. Intravenous glucose is given during episodes of illness. Babies may also be treated with low-protein diet, special infant formula and medical foods. Sometimes, other protein supplements may need to be added to the baby's diet.

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