



## Newborn Screening FACT Sheet

# Glutaric Acidemia, Type II (GA2)

### What is glutaric acidemia, type II?

Glutaric acidemia, type II (GA2) is a condition in which the body is unable to break down certain fats and proteins. It is considered a fatty acid oxidation condition because people affected with GA2 are unable to convert some of the fats and proteins they eat into energy the body needs to function. This causes too many unused fatty acids and other harmful substances to build up in the body. GA2 can cause weak muscle tone, severe heart problems, and death. For some individuals with GA2, detecting it early and beginning treatment may help to prevent some of the severe health outcomes associated with the condition.

### What Causes GA2?

When we eat food, enzymes help break it down. Some enzymes break down fats into their building blocks, called fatty acids. Some enzymes break down amino acids into their building blocks, called amino acids. Other enzymes break down these fatty acids and amino acids for energy. The enzymes electron transfer flavoprotein (ETF) and ETF-ubiquinone oxidoreductase (ETF-QO) helps to break down fats and proteins for energy.

In GA2 one of these enzymes is not working correctly. Only one of the two enzymes needs to be non-working for your baby to have GA2, but it could be either one. If your baby has GA2 his or her body either makes non-working or does not make enough of ETF or ETF-QO.

### What Symptoms or Problems Occur with GA2?

*[Symptoms are something out of the ordinary that a parent notices.]*

Signs of GA2 can begin shortly after birth, in childhood, or even adulthood. Signs of GA2 include:

- sleeping longer or more often
- behavior changes
- irritability
- weak muscle tone (known as hypotonia)
- poor appetite
- fever
- vomiting
- diarrhea
- low blood sugar

Many of these signs may occur when your baby eats foods that his or her body cannot break down. They can be triggered by long periods of time without eating, illnesses, and infections.

If your baby shows any of these signs, be sure to contact your baby's doctor immediately.

### What is the Treatment for GA2?

**1. Dietary Treatments** - Your baby may need to be on a restricted diet in order to avoid certain foods that his or her body cannot break down. A dietician or nutritionist can help you plan a healthy diet for your child. Eating often can also help your baby avoid many of the signs mentioned in the signs and symptoms section.

**2. Medication** - Some children with GA2 take prescription L-carnitine supplements. L-carnitine is a substance naturally produced by the body, but your child's body might not make enough of it. Taking L-carnitine supplements can help the body break down fat for energy and get rid of harmful substances in the body. Your baby's doctor will need to write a prescription for these supplements.

Your baby's doctor might also prescribe riboflavin (a vitamin) or glycine (a building block of proteins) supplements.

### Things to Remember

Children and adults who receive treatment for GA2 can lead healthy lives. Some children who receive treatment may still experience some learning disabilities.

Without treatment, children and adults with GA2 are at risk of liver damage, heart trouble, or brain damage.

Treatment may not be effective for some newborns with GA2. There have been few reported cases of successful GA2 treatment in newborns. Most babies affected with GA2 die in the first few months of life from heart problems.