My Baby Has a Positive Newborn Screening Result: Glutaric Acidemia Type I (GA1)

What is newborn screening?
These are routine tests done soon after birth on every baby born in Manitoba. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including glutaric acidemia type I (GA1).

What does it mean if my baby has a positive newborn screening test for GA1?
This result does not mean that your baby has GA1. It means that more testing is needed because your baby might have GA1. Babies with GA1 are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has GA1.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has GA1 until follow up testing has been done.

What is glutaric acidemia type I (GA1)?
GA1 is a rare inherited (genetic) disease that causes a baby to have problems breaking down certain amino acids (building blocks of protein) called lysine, hydroxylysine and tryptophan. These amino acids are found in most of the foods we eat, including breast milk and infant formula. If the baby cannot break down these amino acids properly, a harmful substance called glutaric acid builds up in the body and can cause serious health problems.

Some people with GA1 also have problems with balance, movement and co-ordination and others may have very mild health problems. There is no way to know who will develop certain problems caused by GA1 and who will not.

What causes GA1?
GA1 happens when an enzyme called glutaryl-CoA dehydrogenase is either missing or not working properly. This enzyme's job is to break down glutaric acid as a source of energy for the body.

How do I find out if my baby actually has GA1?
Blood and urine tests are done to find out if a baby who is screen positive actually has GA1. Sometimes, other tests are done. If you are of Oji-Cree background, additional tests may be able to be completed from the baby’s newborn screening card.

When can my baby have these tests?
Your baby’s doctor or a health care professional at the Winnipeg Children’s Hospital will call you to talk about the results of your baby’s positive newborn screen and arrange more testing as soon as possible.
**Why screen for GA1?**

Babies who have GA1 are usually normal at birth but they are at risk for a metabolic crisis. A metabolic crisis is a serious health condition caused by the build-up of harmful substances in the blood. Symptoms of a metabolic crisis are poor feeding, vomiting, lethargy, excessive sleepiness and irritability. If a metabolic crisis is not treated, breathing problems, seizures, coma, and sometimes death can occur. In GA1, a metabolic crisis may lead to brain damage so the goal of newborn screening for GA1 is to identify an affected newborn before any symptoms might begin and with early treatment and monitoring prevent a metabolic crisis. Such an approach has been proven to help babies and adults with GA1 live healthier lives.

**How is GA1 treated?**

Babies with GA1 are treated and monitored by a team of specialists including a metabolic doctor, metabolic genetic counselor and a dietician. The treatment for GA1 includes a special low protein diet, medical formulas, medications and vitamin supplements may also be given.

**How does a baby get GA1?**

GA1 is an inherited (genetic) disease. A baby with GA1 inherits two non-working copies of the glutaryl-CoA dehydrogenase (GCDH) genes, one copy from each parent. People who have one non-working copy of the GCDH gene are called “carriers.” Carriers of GA1 are healthy and do not have symptoms of GA1.

**Where can I get more information?**

For more information on newborn screening, please talk to your local health care provider or visit the Winnipeg Metabolic Program website [www.wrha.mb.ca/prog/genetics](http://www.wrha.mb.ca/prog/genetics) or the newborn screening section of the Cadham Provincial Laboratory website [www.gov.mb.ca/health/publichealth/cpl/baby.html](http://www.gov.mb.ca/health/publichealth/cpl/baby.html)

For more information on GA1, please visit the Organic Acidemia Association website at [http://www.oaanews.org](http://www.oaanews.org)

**NOTE TO PARENTS/GUARDIANS:** This information is only applicable if your baby has had a positive newborn screening result for glutaric acidemia type I (GA1). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.