INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Glutaric Acidemia Type I (GA 1) Newborn Screening Result

What is newborn screening?

These are routine tests done soon after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including glutaric acidemia type I (GA 1).

What does it mean if my baby has a positive GA 1 newborn screening result?

This result does **not** mean that your baby has GA 1. It means that more testing is needed because your baby **might** have GA 1. Babies with GA 1 are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has GA 1.

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 GA 1 until follow up testing has been done.

What is glutaric acidemia type I (GA 1)?

GA 1 is a rare inherited (genetic) disease that causes a baby to have problems breaking down the amino acids (building blocks of protein) lysine, hydroxylysine and tryptophan. These amino acids are found in most of the foods we eat, including breast milk and infant formula. The body normally breaks lysine and tryptophan down into glutaric acid.

If a baby has GA 1 the body can not break down glutaric acid properly. Glutaric acid and other harmful substances build up in the body and can cause serious health problems. Some people with GA 1 also have problems with balance, movement and co-ordination.

Some people with GA 1 never develop any health problems associated with GA 1. However, there is no way to know who will develop problems caused by GA 1 and who will not.

What causes GA 1?

GA 1 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 has GA 1?

Blood and urine tests are done to find out if a baby who is screen positive actually has GA 1. Sometimes, other tests are done.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange follow up testing as soon as possible.

Why screen for GA 1?
Babies who have GA 1 are usually normal at birth (some may have a bigger head size than average) 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 GA 1, a metabolic crisis may lead to brain damage so the goal of screening for GA 1 is to prevent a metabolic crisis and help people with GA 1 live healthier lives.

How is GA 1 treated?
Babies with GA 1 are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment for GA 1 includes frequent feeding and avoiding fasting (not going a long time without eating). A special low protein, medical formulas, medications and supplements may also be given.

How does a baby get GA 1?
GA 1 is an inherited (genetic) disease. A baby with GA 1 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 GA 1 are healthy and do not have symptoms of GA 1.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit the Parents section of our website at www.newbornscreening.on.ca.

For more information on GA 1, please visit the Organic Acidemia Association website at 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 (GA 1). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

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