



## INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Isovaleric Acidemia 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 isovaleric acidemia (IVA).

### What does it mean if my baby has a positive IVA newborn screening result?

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

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 IVA until follow up testing has been done.

### What is isovaleric acidemia (IVA)?

IVA is a very rare inherited (genetic) disease that causes a baby to have problems breaking down leucine, an amino acid (building block of protein) that is found in most of the foods we eat, including breast milk and infant formula. Leucine is normally broken down into isovaleric acid which is then broken down further as a source of energy for the body.

If the body can not break down isovaleric acid, this acid and other harmful substances build up in the body and can cause serious health problems.

Some people with IVA develop health problems later in infancy or childhood and a few people never have any health problems associated with IVA. There is no way to tell who will have serious problems and who will not.

### What causes IVA?

IVA happens when an enzyme called isovaleryl-CoA dehydrogenase is either missing or not working properly. This enzyme's job is to break down isovaleric acid to use as a source of energy for the body.

### How do I find out if my baby has IVA?

Blood and urine tests are done to find out if a baby who is screen positive actually has IVA.

### 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 IVA?**

Babies who have IVA 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. The goal of screening for IVA is to prevent a metabolic crisis and help people with IVA live healthier lives.

### **How is IVA treated?**

Babies with IVA are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment for IVA includes frequent feeding and avoiding fasting (going a long time without eating). A special low protein diet, medical formulas, medications and supplements may also be given.

### **How does a baby get IVA?**

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

### **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](http://www.newbornscreening.on.ca).

For more information on IVA, please visit the Organic Acidemia Association website at <http://www.oanews.org>.

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

