



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has an Elevated C5OH 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 several organic acid diseases that cause an elevated (higher) level of C5OH.

What does it mean if my baby has an elevated C5OH newborn screening result?

C5OH is a substance that is found in all babies blood and can be measured with newborn screening. An elevated C5OH level does **not** mean that your baby has an organic acid disease. It means that more testing is needed because your baby **might** have an organic acid disease. Babies with organic acid diseases can be healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has an organic acid disease. Sometimes a positive C5OH newborn screen means that the mother has a mild, undiagnosed health problem.

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 an organic acid disease until follow up testing has been done.

What are organic acid diseases?

Organic acid diseases that cause an elevated C5OH are inherited (genetic) diseases that do not allow a baby to break down leucine and isoleucine. These are amino acids (building blocks of protein) that are found in foods that contain protein, including breast milk and infant formula. If a baby cannot break these amino acids down, harmful substances build up in the blood and can cause serious problems with health, growth and learning.

Some people with organic acid diseases never develop health problems and some do not develop problems until childhood or adulthood. However, there is no way to know who will develop problems caused by organic acid diseases and who will not.

What causes organic acid diseases?

Organic acid diseases happen when enzymes needed to break down certain amino acids are either missing or not working properly. The job of these enzymes is to break down amino acids into energy that the body needs to grow and work normally.

How do I find out if my baby has an organic acid disease?

Blood and urine tests are done to find out if a baby who has an elevated level of C5OH actually has an organic acid disease. 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 organic acid diseases?

Babies who have organic acid diseases 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, seizures, coma, and sometimes death can occur. The goal of screening for organic acid diseases is to prevent a metabolic crisis and to help people with organic acid diseases live healthier lives.

How are organic acid diseases treated?

Babies with organic acid diseases are treated and monitored by a team of specialists including a metabolic genetics doctor. Babies with organic acid diseases should not go a long time without eating. Depending on which organic acid disease a baby has, special diets low in protein, medical formulas, medications and supplements may be recommended.

How does a baby get an organic acid disease?

Organic acid diseases are inherited (genetic) diseases. A baby with an organic acid disease inherits two non-working copies of a gene that codes for an enzyme needed to break down an amino acid, one copy from each parent. People who have one non-working copy of the gene are called “carriers.” Carriers of organic acid diseases are healthy and do not have symptoms of organic acid diseases.

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 organic acid diseases, please visit the Organic Acidemia Association website at www.oaanews.org.

NOTE TO PARENTS/GUARDIANS: This information is only applicable if your baby has had a positive newborn screening result with elevated C5OH. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

