



INFORMATION FOR PARENTS/GUARDIANS: My Baby Has a Positive Maple Syrup Urine Disease 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 maple syrup urine disease (MSUD).

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

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

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 MSUD until follow up testing has been done.

What is maple syrup urine disease (MSUD)?

MSUD is a very rare inherited (genetic) disease that causes a baby to have problems breaking down leucine, isoleucine and valine. These are amino acids (building blocks of protein) that are found in most of the foods we eat, including breast milk and infant formula.

If the body can not break down leucine, isoleucine and valine, these amino acids and other harmful substances build up in the body and can cause serious health problems. The disease got its name because the urine of affected babies may smell like maple sugar or burnt sugar.

Some people with MSUD develop health problems later in infancy or childhood but newborn screening cannot tell who will have problems as a newborn and who will not.

What causes MSUD?

MSUD happens when an enzyme called branched chain ketoacid dehydrogenase (BCKAD) is either missing or not working properly. This enzyme's job is to break down leucine, isoleucine and valine to use as a source of energy for the body.

How do I find out if my baby has MSUD?

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

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 MSUD?

Babies who have MSUD 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 MSUD is to prevent a metabolic crisis and help people with MSUD live healthier lives.

How is MSUD treated?

Babies with MSUD are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment for MSUD may include a special low protein diet, medical formulas, medications and supplements.

How does a baby get MSUD?

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

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 MSUD, please visit the MSUD Family Support Group website at <http://www.msud-support.org>.

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

