



INFORMATION FOR PARENTS/GUARDIANS:

My Baby Has a Positive Propionic Acidemia/Methylmalonic 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 propionic acidemia (PA) and methylmalonic acidemia (MMA).

What does it mean if my baby has a positive PA/MMA newborn screening result?

This result does not mean that your baby has PA or MMA. It means that more testing is needed because your baby might have PA or MMA. Babies with PA and MMA are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has PA or MMA. Sometimes a positive PA/MMA newborn screen means that the mother has low levels of vitamin B12.

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 PA or MMA until follow up testing has been done.

What are propionic acidemia (PA) and methylmalonic acidemia (MMA)?

PA and MMA are two different inherited (genetic) conditions that both cause a baby to have problems breaking down isoleucine, valine, methionine and threonine. These are amino acids (building blocks of protein) and are found in many of the foods we eat, including breast milk and infant formula. If the body can not break down these amino acids, harmful substances build up and can cause mental retardation, organ failure and other serious health problems.

Some people with PA or MMA do not develop problems until childhood or adulthood or have a milder form of these diseases. However, newborn screening cannot tell who will develop problems caused by PA or MMA early in life and who will not.

PA affects about 1 in every 100 000 babies born in Ontario. MMA affects about 1 in every 50 000 babies in Ontario.

What causes PA and MMA?

PA happens when an enzyme called propionyl CoA carboxylase (PCC) is either missing or not working properly. MMA happens when an enzyme called methylmalonyl-CoA mutase (MUT) is either missing or not working properly or when the body has problems using vitamin B12 (an important co-factor or helper of the MUT enzyme). The job of these enzymes is to break down certain amino acids to use as a source of energy for the body.

How do I find out if my baby has PA or MMA?

Blood and urine tests are done to find out if a baby who is screen positive actually has PA or MMA. 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 PA and MMA?

Babies who have PA or MMA 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. When PA and MMA are diagnosed and treated early, many other serious health problems may also be prevented. The goal of screening for PA and MMA is to avoid a metabolic crisis and help people with PA and MMA live healthier lives.

How are PA and MMA treated?

Babies with PA and MMA are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment can include a special low protein diet, medical formulas, medications and supplements. Babies with PA and MMA are monitored often for their amino acid levels, development and for other health issues associated with PA and MMA.

How does a baby get PA or MMA?

PA and MMA are inherited (genetic) diseases. A baby with PA inherits two non-working copies of the propionyl CoA carboxylase (PCC) genes, one copy from each parent while a baby with MMA inherits two non-working copies of the methylmalonyl-CoA mutase (MUT) genes or two non-working copies of the genes that process vitamin B12, one from each parent. People who have one non-working copy of the PCC gene, the MUT gene or the cofactor genes are called “carriers.” Carriers of PA and MMA are healthy and do not have symptoms of PA or MMA.

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 PA and MMA, 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 for propionic acidemia (PA) or methylmalonic acidemia (MMA). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

