

# NEWBORN SCREENING BC

## Blood spot card screening | Parent information sheet

### Methylmalonic Acidemia (MMA)

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This is done to identify a number of rare but treatable conditions which usually do not have any symptoms early on. Knowing if your child has one of these conditions early in life is important, because early diagnosis allows for early treatment and better health outcomes.

Your baby had a positive newborn screen result for Methylmalonic Acidemia (MMA). **This result does not mean that your baby has MMA but means the chance is increased and more testing is needed.**

#### What is MMA?

MMA is a group of inherited conditions. Babies with MMA cannot properly break down certain amino acids (the building blocks of protein) and certain types of fat. Because of this, too much methylmalonic acid and other harmful substances build up in the body. This buildup can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screen measures the amount propionylcarnitine and methylmalonic acid which are increased in babies with MMA. Your baby's screen showed a higher-than-expected level.

#### What are the signs and symptoms of MMA?

Babies with MMA are usually healthy at birth. But without treatment, the buildup of harmful substances put the baby at risk of having an episode of metabolic crisis. Signs of **metabolic crisis** include vomiting, dehydration, lethargy (very sleepy), poor feeding, and floppiness. If severe and left untreated, delays in development and loss of consciousness can occur.

How a baby with MMA presents can vary. Some babies are only mildly affected and do not have symptoms but may still

be at risk for an episode of metabolic crisis. A metabolic crisis can be triggered by events such as fasting, fever, or illness.

#### What causes MMA?

In most cases of MMA, an enzyme called **methylmalonyl-CoA mutase (MUT)** is missing or not made properly. This enzyme is made by a gene called MMUT. A gene is a set of instructions on how to make a protein or enzyme. Changes in a gene, called disease-causing variants result in little or no functioning enzyme being made.

We all have two copies of the MMUT gene. A baby born with MMA has received (inherited) two non-working copies of the gene, one from each parent. Parents of a child with MMA are usually carriers. A carrier is a person with one working and one non-working copy of a gene. People usually don't know they are a carrier because carriers are unaffected.

The MUT enzyme needs vitamin B12 to work. As a result, some cases of MMA are caused by low vitamin B12 levels in mom or a problem in how the body processes vitamin B12 (Cobalamin disorder A,B).

#### What happens next?

Your primary care provider will review more information with you. If needed, a specialist doctor in the Biochemical Genetics Clinical Service (BGCS) will contact you to answer questions and organize a visit for your family at BC Children's Hospital in Vancouver.

The doctor will meet your baby and discuss with you further testing options to confirm if your baby has MMA. Testing may include a repeat newborn screen, urine organic acids and genetic testing.

#### How is MMA treated?

The goal of treatment is to try to prevent an episode of metabolic crisis. Babies with MMA must not go a long time

without eating. Dietitians and specialists will support families as they learn about the required dietary considerations and measures to avoid fasting. Children may need to go to the hospital when sick with a cold or flu if they are unable to eat and drink. In addition, a low protein diet, special formula, supplements, and medications may be recommended.

Treatment is coordinated by specialists at BC Children's Hospital.

### **What is the outcome of treatment?**

Treatment helps to prevent episodes of metabolic crisis and other symptoms and gives babies the best chance possible for normal growth and development.

### **How do you feel?**

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. If you are feeling overwhelmed or have questions, you can talk with your health care provider to discuss supports available.

### **Where can I get more information?**

Talk to your primary care provider. You may also call the BGCS Newborn Screening Team at 604-875-2623.

Information about the BC Newborn Screening Program can be found at:

[www.newbornscreeningbc.ca](http://www.newbornscreeningbc.ca)

Information about MMA can be found at:

- [www.newbornscreening.info/Parents/organicac-iddisorders/MMA.html](http://www.newbornscreening.info/Parents/organicac-iddisorders/MMA.html)
- [www.oaanews.org](http://www.oaanews.org)

*This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis or treatment. Always talk to your health care provider about specific health concerns.*