NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Medium Chain Acyl-CoA dehydrogenase deficiency (MCAD)

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This screen tests for a number of rare conditions. 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 MCAD deficiency. This result does not mean that your baby has MCAD deficiency but means the chance is increased and more testing is needed.

What is MCAD deficiency?

Our body breakdowns the sugars, fats, and proteins we eat to make energy. Our cells need this energy to work properly. The main energy source is usually sugar (carbohydrates). But during long periods of time without food (fasting), our body relies more heavily on stored fat for energy. People who have MCAD deficiency cannot breakdown certain fats called **medium chain fatty acids** to make energy. When fasting (such as during a cold or flu) babies with MCAD deficiency do not make enough energy for proper cell function and can become very ill.

What are the signs and symptoms of MCAD deficiency?

Babies with MCAD deficiency are usually healthy at birth and remain healthy if they do not fast. During fasting episodes, a child may have a **metabolic crisis**, which presents as lethargy (very sleepy and unresponsive), floppiness, vomiting, seizures, and low levels of glucose and ketones in their blood. If untreated, this can progress quickly to a coma and loss of life.

What causes MCAD deficiency?

The enzyme needed to breakdown medium chain fatty acids is missing or not work properly in people affected with MCAD deficiency. The MCAD enzyme is made by a gene called the ACADM gene.

A gene is a set of instructions (like a recipe) on how to make a protein or enzyme. Changes in the ACADM gene, called disease causing variants result in the enzyme not working properly or not being made at all.

We all have two copies of the ACADM gene. A person with one working and one non-working ACADM gene is called a carrier. People usually don't know they are a carrier because carriers are unaffected. A baby born with MCAD deficiency has received a non-working ACADM gene from both parents.

What happens next?

A specialist in the Biochemical Genetics Clinical Service (BGCS) team will contact you to provide more information, answer questions, and organize a visit for your family at BC Children's Hospital in Vancouver.

The specialist will examine your baby and discuss with you further testing options to confirm if your baby has MCAD. Testing may include a repeat newborn screen and genetic testing of the ACADM gene.

How is MCAD deficiency treated?

Frequent feeds ensures that a child with MCAD deficiency does not undergo any prolonged period of fasting. To prevent a metabolic crisis, babies with MCAD must not go a long time without eating. Children with MCAD may need to go to the hospital when sick with colds or flu if they are unable to eat and drink.

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

What is the outcome of treatment?

Treatment is very effective at preventing metabolic crises. With early treatment and careful monitoring most babies with MCAD will lead healthy lives with normal growth and development. As children get older, the risk of metabolic crisis decreases.

What is its incidence?

MCAD deficiency affects about 1 in every 12,000 babies born in BC.

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 doctor or midwife. 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

Information about MCAD deficiency can be found at:

- **http://www.fodsupport.org/**
- **http://www.geneclinics.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.