NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Carnitine Uptake Deficiency (CUD)

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 Carnitine Uptake Deficiency (CUD). **This result does not mean that your baby has CUD but means the chance is increased and more testing is needed.**

What is CUD?

CUD is a rare inherited condition. It is a type of fatty acid disorder which means people with CUD cannot breakdown fats (fatty acids) to make energy. Our bodies normally breakdown 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 on stored fats. When fasting (such as during a cold or flu), babies with CUD do not make enough energy for proper cell function and can become very ill.

A protein called ***carnitine transporter protein*** moves ***carnitine*** into our cells where it plays an essential role in the process of turning fat into energy. People with CUD have low levels of carnitine.

The newborn screen measures the amount of carnitine. Your baby's screen showed a lower-than-expected level.

What are the signs and symptoms of CUD?

Babies with CUD are usually healthy at birth. However, without treatment, a child with CUD is at risk of having an episode of low blood sugar, lethargy (very sleepy and unresponsive), poor feeding and liver problems. These episodes are often triggered by events such as fever or illness.

Symptoms of CUD are variable with some people not being identified until later in life. Some older children can develop muscle pain and heart problems.

What causes CUD?

The carnitine transporter protein is missing or not working properly in people affected with CUD. This transporter protein is made by a gene called SLC22A5. A gene is a set of instructions on how to make a protein. Changes in a gene, called disease-causing variants result in little or no functioning protein being made.

We all have two copies of the SLC22A5 gene. A baby born with CUD has received (inherited) two non-working copies of the SLC22A5 gene, one from each parent. Parents of a child with CUD are usually carriers. A carrier is a person with one working and one non-working copy of the gene. People usually don't know they are a carrier because carriers are unaffected.

What happens next?

Your primary care provider will review more information with you and arrange a repeat newborn screen on the baby to confirm the result. They will also arrange an acylcarnitine bloodspot test on the baby's mother to assess her carnitine level. This is because sometimes the low carnitine level picked up by the newborn screen is from an undiagnosed mother with CUD.

If the repeat results show a normal carnitine level in the baby and mother, then there is no evidence of CUD. This is a reassuring result and no further follow up is needed.

If the repeat results show a low carnitine level in the mother or baby, then a referral to a specialist doctor in the Biochemical Genetics Clinical Service (BGCS) team will be made. They will contact you to provide more information, answer questions, and organize a visit for your family at BC Children's Hospital in Vancouver.

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.

The doctor will meet your baby and discuss with the family further testing options to confirm if the baby or mother has CUD. Testing may include a special blood and urine test and genetic testing.

How is CUD treated?

The main treatment for CUD is oral carnitine supplementation and to avoid fasting.

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

What is the outcome of treatment?

If treatment is started early, children with CUD have a good prognosis.

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

Information about CUD can be found at:

- **www.newbornscreening.info/Parents/fattyaciddisorders/Carnitine.html**
- **https://rarediseases.org/rare-diseases/systemic-primary-carnitine-deficiency**

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.