NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Citrullinemia (CIT) / Argininosuccinic Aciduria (ASA)

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 which may suggest either Citrullinemia (CIT) or Argininosuccinic Aciduria (ASA). **This result does not mean that your baby has one of these conditions but means the chance is increased and more testing is needed.**

What is CIT/ASA?

CIT/ASA are both rare inherited disorders. They occur when the body's normal process to remove ammonia called the ***urea cycle*** is not working properly. As a result, too much ammonia (***hyperammonemia)*** and other harmful substances build up in the blood and body. Our body makes ammonia when proteins are broken down. Normally the ammonia is turned into urea which is harmless and easily removed from the body through urine. High levels of ammonia can cause serious health problems if it is not diagnosed and treated early.

The newborn screen test measures the level of citrulline which is elevated in babies with CIT/ASA. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of CIT/ASA?

Babies with CIT/ASA are usually healthy at birth. However, without treatment, high levels of ammonia can build up. Signs of hyperammonemia include lethargy (very sleepy and unresponsive), poor feeding, floppiness and vomiting. Episodes of hyperammonemia can be triggered by events such as fasting, fever, illness, and after a high protein meal.

Most of the time, symptoms present in infancy, but some people have a milder form and do not have symptoms until childhood.

What causes CIT/ASA?

One of the enzymes needed to remove ammonia from the body is not working properly in people affected with CIT/ASA. For CIT, the enzyme is called ***argininosuccinate synthase 1 (ASS1)***. For ASA, the enzyme is called ***argininosuccinate lyase (ASL)***. These enzymes are made in our cells by genes called ASS1 and ASL. 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 ASS1 and ASL genes. A baby born with CIT has received (inherited) two non-working copies of the ASS1 gene, one from each parent. A baby born with ASA has inherited two non-working copies of the ASL gene, one from each parent. Parents of a child with CIT/ASA are usually carriers. A carrier is a person with one working and one non-working copy of the gene involved. 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. 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 CIT/ASA. Testing may include a repeat newborn screen, a blood test to measure plasma amnio acids, and genetic testing.

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.

How is CIT/ASA treated?

A special low protein diet is prescribed under the care of a dietitian and specialist along with supplements and medications as needed. In addition, babies with CIT/ASA must not go a long time without eating. This is because during a fast their body will breakdown its own protein as a source of energy to meet its energy demands. So, frequent feeds are recommended. Children may need to go to the hospital when sick with a cold 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?

If diagnosed early, treatment can help to prevent these health problems and allow children with these diseases to lead the healthiest lives possible. Some children can still have high ammonia levels with treatment, especially during times of illness, and require close monitoring and follow up.

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 CIT/ASA can be found at:

- **www.newbornscreening.info/Parents/aminoaciddisorders/ASAS.html**
- **www.newbornscreening.info/Parents/aminoaciddisorders/ASAL.html**
- **www.nucdf.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.