NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Congenital Adrenal Hyperplasia (CAH)

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 Congenital Adrenal Hyperplasia (CAH). **This result does not mean that your baby has CAH but means the chance is increased and more testing is needed.**

What is CAH?

CAH is an inherited condition in which the adrenal gland cannot make important hormones called ***cortisol*** and ***aldosterone***. Hormones are chemical messengers used by the body to control body functions. Cortisol is important in helping the body cope with physical stress, like illness or injury. Aldosterone is important for controlling salt balance.

Babies who do not make enough of these hormones are not able to properly control the amount of salt and water in their body. This can lead to a ***salt-losing crisis,*** which if untreated could result in loss of life. The hormone imbalance can also result in too much male hormone being made.

The newborn screen test measures the level of ***17-OHP*** (17-hydroxyprogesterone) and related markers. These are increased in babies with CAH. Your baby's screen showed higher-than-expected levels.

What are the signs and symptoms of CAH?

Some female babies and all male babies with CAH appear normal at birth but can be at risk for a salt-losing crisis in the first few weeks of life. These babies may present with failure to thrive (poor growth), vomiting and severe dehydration with electrolyte imbalance (low sodium and high potassium in the blood).

Because of the possibility of too much male hormone being made during the pregnancy, some female babies may have male-type changes in the appearance of their external genitals, such as enlargement of the clitoris**.**

What causes CAH?

One of the enzymes needed to make cortisol and aldosterone is not working properly in people with CAH. The most common cause of CAH is deficiency of an enzyme called ***21-hydroxylase (21-OH***). Not enough of this enzyme causes high levels of 17-OHP.

The 21-OH enzyme is made by a gene called CYP21A2**.** A gene is a set of instructions on how to make a protein or enzyme. Changes in the CYP21A2 gene, called disease causing variants result in little or no functioning enzyme being made.

We all have two copies of the gene. A baby born with 21-OH deficiency has received (inherited) two non-working copies of the CYP21A2 gene, one from each parent. Parents of a child with 21-OH deficiency 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. If needed, a specialist doctor in endocrinology will contact you to answer questions and organize a visit for your family.

The doctor will meet your baby and discuss with you further testing options to confirm if your baby has CAH. Testing includes measuring 17-OHP and other adrenal hormones in the blood.

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 CAH treated?

Replacement of deficient hormones is an effective means of preventing a salt-wasting crisis and preventing long-term complications.

Treatment is coordinated by a specialist doctor in endocrinology.

What is the outcome of treatment?

Infants who are identified early and treated appropriately have a very good prognosis but require lifelong management and monitoring.

What is the incidence of CAH?

CAH affects about 1 in every 15,000 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 primary care provider.

Information about the BC Newborn Screening Program can be found at
www.newbornscreeningbc.ca

Information about CAH can be found at:

- **http://www.caresfoundation.org/**
- **http://www.bcchildrens.ca/endocrinology-diabetes-site/documents/cahbooklet.single.pdf**
- **https://www.mayoclinic.org/diseases-conditions/congenital-adrenal-hyperplasia/symptoms-causes/syc-20355205**

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.

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