

## BC Newborn Screening Program

## Information Sheet

### Congenital Adrenal Hyperplasia (CAH) – Endocrine Disorder

#### What is Congenital Adrenal Hyperplasia (CAH)?

Congenital Adrenal Hyperplasia (CAH) is an inherited defect in which the adrenal gland has an impaired ability to make cortisol and aldosterone due to a mutation in one of the enzymes required for their synthesis. Cortisol is important in coping with physical stress and aldosterone is important for controlling salt balance. The underproduction of these hormones may mean that an infant cannot regulate salt and fluids, leading to a salt-losing crisis and possibly death. In addition, because of the block in certain hormonal pathways, excess male hormones are produced.

#### What is its incidence?

CAH affects about 1 in every 16,000 babies born in BC.

#### What causes the disease?

The most common cause of CAH is deficiency of 21-hydroxylase (21-OH) enzyme, a crucial enzyme in cortisol and aldosterone synthesis. Levels of hormone precursors such as 17-hydroxyprogesterone and androgens are elevated.

#### What are the clinical features of the disease?

CAH is different from many of the other conditions in the newborn screen in that girls may be symptomatic at birth. With the overproduction of male hormones during pregnancy, some chromosomally female babies have masculinization of the external genitalia to varying degrees. Some female babies and all male babies appear normal at birth but can be at risk for a salt-losing crisis.

These babies may present with failure to thrive, vomiting, dehydration, hypotension, hyponatremia, hyperkalemia, and may go into shock and die. This crisis occurs within the first few weeks of life. In the long term, individuals with CAH may continue to have an imbalance in certain hormones, leading to symptoms such as precocious puberty, short stature, or reduced fertility.

#### How is the diagnosis confirmed?

The diagnosis of CAH is confirmed by measuring 17-OHP and other adrenal hormones in the blood.

Mutation analysis of the CYP21A2 gene is done once the diagnosis is established.

Diagnostic testing is arranged by specialists at BC Children's Hospital.

#### What is the treatment of the disease?

Replacement of deficient hormones is an effective means of preventing a salt-wasting crisis and preventing long-term complications as indicated above. Parents of female babies who have had virilization may opt for surgery to improve the appearance of the external genitalia.

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

#### What is the outcome of treatment?

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

## Can a family have more than one child with CAH?

CAH is inherited as an autosomal recessive disease. Parents of a child with CAH are assumed to be carriers for the disease and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with the condition. Women who have had a previous child with CAH are often prescribed dexamethasone in pregnancy in order to prevent virilization of a female fetus. Prenatal testing for CAH can be done as early as 10-12 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended. Unaffected siblings of a child with CAH have a 2/3 chance of being carriers. CAH carriers are healthy and do not have symptoms of CAH.

### Resources

<http://www.congenitaladrenalyhyperplasia.org/>  
<http://www.caresfoundation.org/>  
<http://www.medhelp.org/nadf/diseases/cah.htm>  
<http://www.hopkinschildrens.org/specialties/categorypages/cah/index.html>  
<http://www.sickkids.ca/childphysiology/cpwp/Genital/GenitalCAH.htm>  
[http://www.rch.org.au/cah\\_book/index.cfm?doc\\_id=1375](http://www.rch.org.au/cah_book/index.cfm?doc_id=1375)

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