Alberta Health Services

Newborn Metabolic Screening

Information for Health Professionals

Congenital Adrenal Hyperplasia (CAH)
(endocrine condition)

Also known as:

- adrenogenital syndrome (AG syndrome)
- 21-hydroxylase deficiency

What is CAH?

CAH is a condition in which enzymes required for the synthesis of cortisol and aldosterone by the adrenal gland are missing. Cortisol and aldosterone are important in salt and water balance in the body and cortisol is involved in the body's normal response to stress, for example, during an infection. There may be an increase in androgens (male hormones) secreted by the adrenal gland.

What causes CAH?

The most common cause of CAH is a decreased or absent activity of steroid 21-hydroxylase, an enzyme required for cortisol and aldosterone synthesis. This results from mutations in the CYP21 gene. There are several different mutations in the gene which result in either mild or severe forms of CAH.

How common is CAH?

The incidence of CAH is about 1 in every 15,000 infants born in Canada.

What are the clinical features of CAH?

Male infants appear normal at birth. Female infants may have masculinization of the external genitalia because of an excess of male adrenal androgens. Both males and females with severe CAH are at risk for salt-wasting crisis. Infants may present with failure to thrive, vomiting, dehydration, hyponatremia, hyperkalemia and shock in the first few weeks of life.

Milder forms of CAH present later in life and may not be detected on the newborn blood spot screen.

What is the screening test for CAH?

An increased 17-hydroxyprogesterone (17OHP) concentration is detected on the newborn blood spot screen. Newborn blood spot screening is not 100% perfect. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of CAH is confirmed by measurement of serum 17OHP and other adrenal hormones together with mutation analysis of the CYP21 gene. Specialists at the clinics listed below will arrange diagnostic testing.

How is CAH treated?

Cortisol and aldosterone replacement prevents salt-losing crisis and decreases the buildup of adrenal androgens. Parent education is an important component of treatment, including advising parents on actions to be taken when their child develops a minor illness. Lifelong hormone replacement is necessary. Surgery may be required for female infants to correct masculinization of the external genitalia.

What is the outcome of treatment for CAH?

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

Is CAH inherited?

CAH is inherited as an autosomal recessive trait. Parents of a child with CAH are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. CAH carriers are healthy. Genetic counselling and prenatal testing is available to all families. Treatment of the mother with steroids during pregnancy may prevent masculinization in a female fetus.

For additional resources, please call:

Pediatric Endocrinology
Stollery Children's Hospital
1C4, 8440 – 112 St. NW
Edmonton, AB T6G 2B7
Phone: 780-407-8249
Fax: 780-407-1509

Emergency consultations:
Phone 780-407-8822 and ask for the specialist on call for pediatric endocrinology.

Pediatric Endocrine Clinic
Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7003
Fax: 403-955-7639

Emergency consultations:
Phone 403-955-7211 and ask for the specialist on call for endocrine diseases.