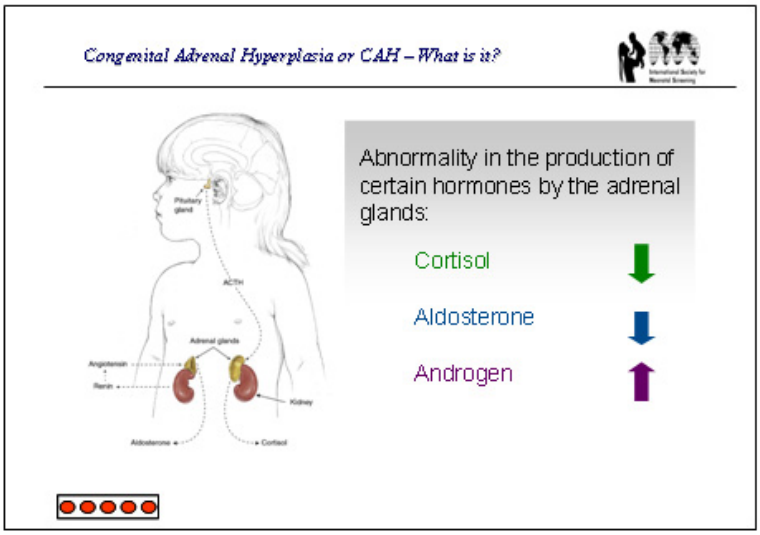
Congenital Adrenal Hyperplasia (CAH)
Information for the parents/carers

What is congenital adrenal hyperplasia?



A person with congenital adrenal hyperplasia (or CAH) doesn't make enough of the hormones cortisol and aldosterone, and make too much androgen, which is a male sex hormone. If you have CAH, your adrenal gland tries to make cortisol and aldosterone, but it can't make enough. CAH is usually diagnosed at birth or in early childhood.

What are the symptoms of CAH?

Female children with severe CAH might be born with ambiguous genitalia, which means that the genitals may look more male than female. As they get older, girls with CAH may develop facial hair and a deep voice, and they may have abnormal menstrual periods or no periods at all. Boys with CAH often have well-developed muscles and early development of masculine features.

People with CAH may be shorter than most average adults. They may have acne and blood pressure problems. When they get colds and sinus infections they don't get better as quickly as other people do. Women with mild CAH often have irregular periods. They may have trouble getting pregnant.

Salt-Wasting CAH

A Severe form of CAH is called " Salt-Wasting" CAH. A baby with this form of CAH loses large amounts of salt (sodium) in the urine. This can lead to symptoms such as:

- Dehydration
- Muscle weakness
- Poor growth
- Vomiting
- Weakness of the heart

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

How does CAH occur?

CAH is a genetic disorder. Parents of an affected child carry a genetic trait causing CAH. Both parents pass the trait to a child with CAH. There is a 1 in 4 chance that each child will have CAH when both parents carry the trait for the disorder

If I have CAH, will my children get it?

If you and your partner have any form of CAH, your children might also have it. Remember, some people can have mild CAH and not know it, so you should tell your doctor as soon as you think you or your partner may be pregnant. A baby can be treated before he or she is born if the mother takes the medicine. If your baby has CAH, treatment should begin as soon as you know you are pregnant.

How is CAH diagnosed?

In neonates, serum levels of 17-hydroxyprogesterone are measured in the dried blood on filter paper of the newborn screen. If levels are elevated compared with age-adjusted norms, the diagnosis must be confirmed by identifying low blood levels of deoxycortisol, cortisol, deoxycorticosterone, corticosterone, progesterone, and 17-hydroxyprogesterone and by identifying high levels of DHEA and androstenedione, measured in whole blood from the neonate.

If you are pregnant or trying to get pregnant and there is a history of CAH in the family, talk to your doctor about genetic testing. Your doctor may order a blood test if there are signs that your infant or child has CAH.

What treatment is available for CAH?

Right now, there is no cure for CAH, but there is treatment. Your doctor will prescribe a form of cortisol that will need to be taken every day. The goal of treatment is to get the hormones to a normal level. Extra cortisol may need to be taken during times of stress, such as surgery.

Treatment:

The goal of treatment is to return hormone levels to normal. This is done by taking a form of cortisol (dexamethasone, fludrocortisone, or hydrocortisone) every day. Additional doses of medicine are needed during times of stress, such as severe illness or surgery.

The gender of a baby with ambiguous genitalia is determined by examination of the chromosomes (karyotyping). Reconstructive surgery for girls with masculine external genitalia is usually performed between the ages of 1 and 3 months to correct the abnormal appearance.

Parents of children with congenital adrenal hyperplasia should be aware of the side effects of steroid therapy. They should report signs of infection and stress to their health care provider

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm

because increases in medication may be required. In addition, steroid medications cannot be stopped suddenly, or adrenal insufficiency will result.

Expectations (prognosis):

The outcome is usually associated with good health, but short stature may result even with treatment. Males have normal fertility. Females may have a smaller opening of the vagina and lower fertility.

Medication to treat this disorder must be continued for life.

Complications:

- Adrenal crisis , including hyponatremia and shock (especially in newborns)
- Abnormal female external genitalia (internal organs are normal)
- Early development of male sexual characteristics
- Short adult stature despite early, rapid childhood growth
- Tumors of the testes in adult men
- High blood pressure
- Low blood sugar
- Side effects of corticosteroids used as treatment

Calling your health care provider:

Call for an appointment with your health care provider if your child develops symptoms of this disorder.

Also, if you had a child with this disorder or you have a family history of this disease and you plan to have other children, you should discuss this with your health care provider before conceiving a child. Genetic counseling is important if there is a history of congenital adrenal hyperplasia.

Support Groups:

National Adrenal Diseases Foundation: www.medhelp.org/nadf/

CARES (Congenital Adrenal hyperplasia Research, Education, and Support) Foundation: www.caresfoundation.com

The MAGIC Foundation: www.magicfoundation.org

Tel: 018-299 2310/293 9510 | Fax: 018-299 2316 | E-mail: NBS@Ampath.co.za
Address: Biochemistry, NWU, Private Bag X6001, Potchefstroom, 2520
www.newbornscreening.co.za OR www.ampath.co.za/newborn_screeningIndex.htm