Newborn Screening Programme for
Inborn Errors of Metabolism
Information leaflet series (No. 22)

Congenital Adrenal Hyperplasia

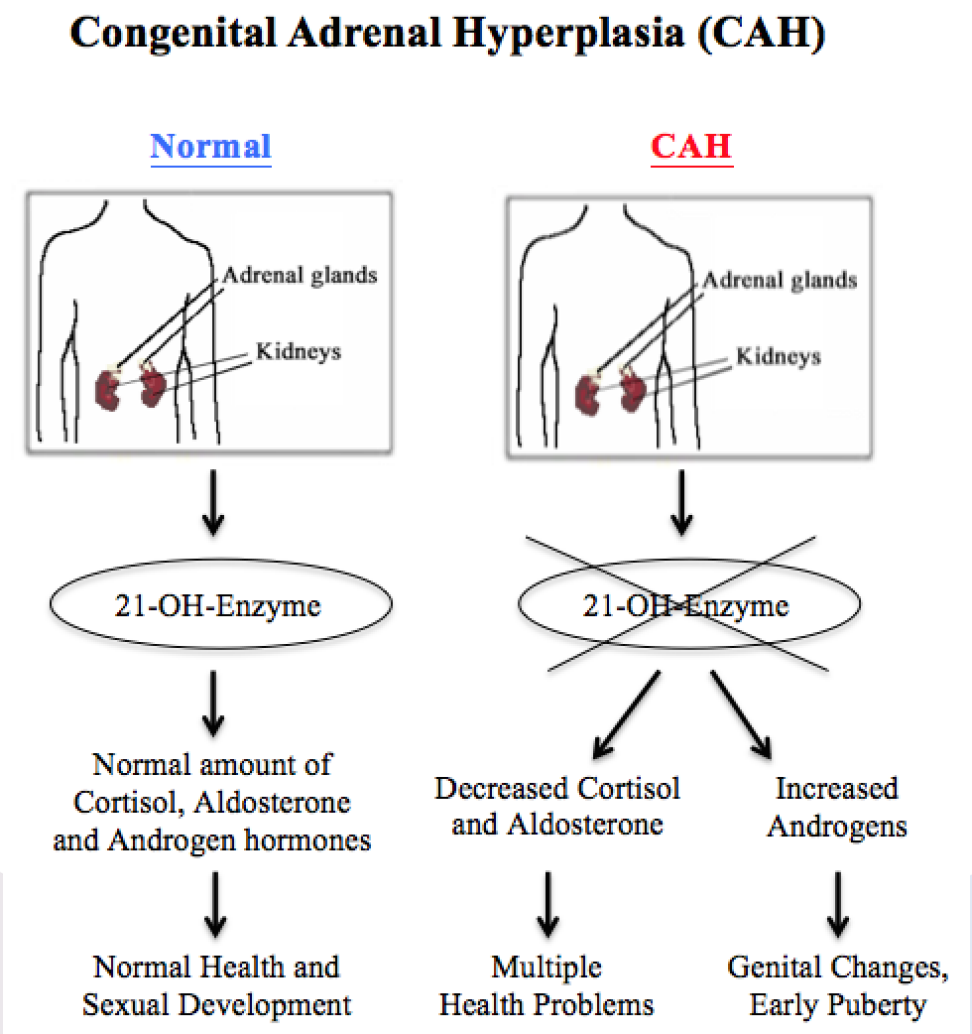
For general queries on Newborn Screening Programme for Inborn Errors of Metabolism, please call: ☎ 5741 4280 (Department of Clinical Genetics, Hospital Authority)

醫院管理局
HOSPITAL AUTHORITY

What is Congenital Adrenal Hyperplasia (CAH)?

Congenital adrenal hyperplasia (CAH) affects the adrenal glands. The adrenal glands make different hormones, such as cortisol (the body's natural steroid which helps the body deal with stress and illness), aldosterone (helps regulate salt and water levels) and androgens (male sex hormones).

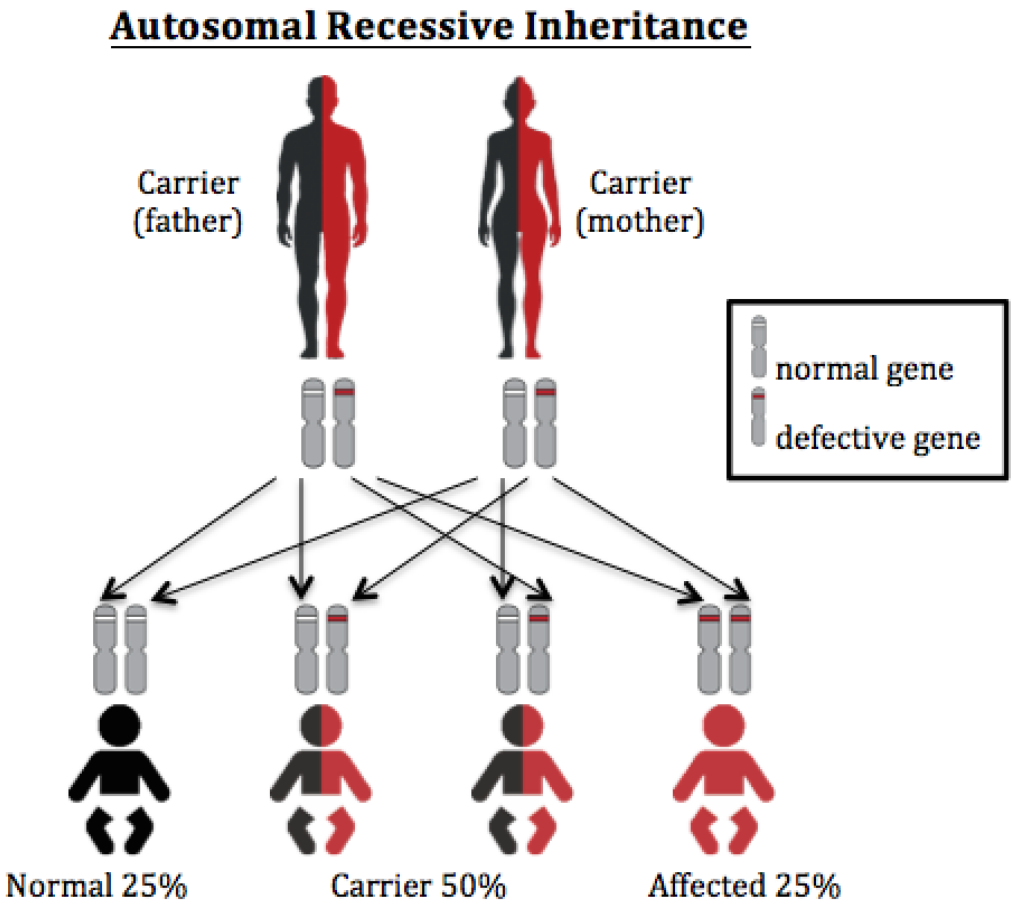
CAH occurs when an enzyme called "21-hydroxylase" (21-OH) is either absent or not functioning properly. A person who has CAH does not make enough cortisol and aldosterone, and makes too many androgens. Lacking cortisol and aldosterone means that the body is less able to cope with stress, and can be life-threatening. Too much androgens cause male characteristics to appear early in boys, or inappropriately in girls.



How is CAH inherited?

Everybody has two copies of genes, one from each parent, which tell the body how to make specific enzymes.

Congenital adrenal hyperplasia is an autosomal recessive disease. Only when babies inherit two faulty copies of the gene for congenital adrenal hyperplasia from parents, the enzyme made does not work properly or is not even made at all.



What may happen if your baby has CAH?

Each baby with CAH may have different clinical presentations. There are three main forms of CAH, which includes:

1. Salt-wasting classic CAH
2. Simple virilising form
3. Non-classical CAH

Early Signs and Symptoms of salt-wasting classic CAH

- Poor feeding
- Irritability and drowsiness
- Repeated vomiting
- Weight loss
- Dehydration
- Collapse and coma

Both salt-wasting classic CAH and simple virilising CAH may present at birth, as excessive male hormones are made by the fetus and causes the genitals of female fetuses to develop male-like features.

Non-classical CAH can manifest in childhood or later in life with symptoms of excessive male hormones such as early puberty, acne, excess hair growth, and menstrual irregularities.

What is the treatment for CAH?

Babies with CAH benefit significantly from early treatment and can have active lives. Children with CAH are cared for by a multidisciplinary team which includes doctors who specialized in hormones and surgeons.

Oral medications to replace and mimic the body's normal cortisol and aldosterone levels and will be required lifelong. Some females with CAH may require surgery to correct their masculinized genitalia.

Cortisol is a stress hormone and is increased naturally when the body is under period of stress, such as during illness and surgery. In children with CAH, their bodies cannot produce cortisol adequately, therefore replacement oral medications will have to be increased during illness. Your doctor will advise you what to do when your child is sick. It is very important to work out with your doctor a care plan.

When should I seek immediate help? What should I do?

If you are worried that your baby is ill, it is important to follow medical advice. Bring your baby to your local accident and emergency department immediately. Take any information that you have been given about CAH, including this pamphlet, to the hospital with you.