Newborn Metabolic Screening Program

CUHK-BCM Joint Centre for Medical Genetics
The Chinese University of Hong Kong

Information for Parents
Screening for

Congenital Adrenal Hyperplasia

(CAH)

Enquiries:
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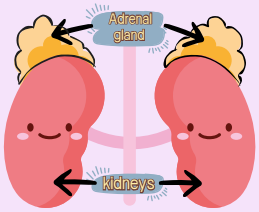
website:

If you wish to join this screening program, please contact your obstetrician during antenatal period or contact your paediatrician within 7 days after delivery

What are the functions of adrenal glands?

Adrenal glands are a pair of small organs located above the kidneys, which produce the correct amount of hormones including cortisol, mineralocorticoids, androgens, and adrenaline.

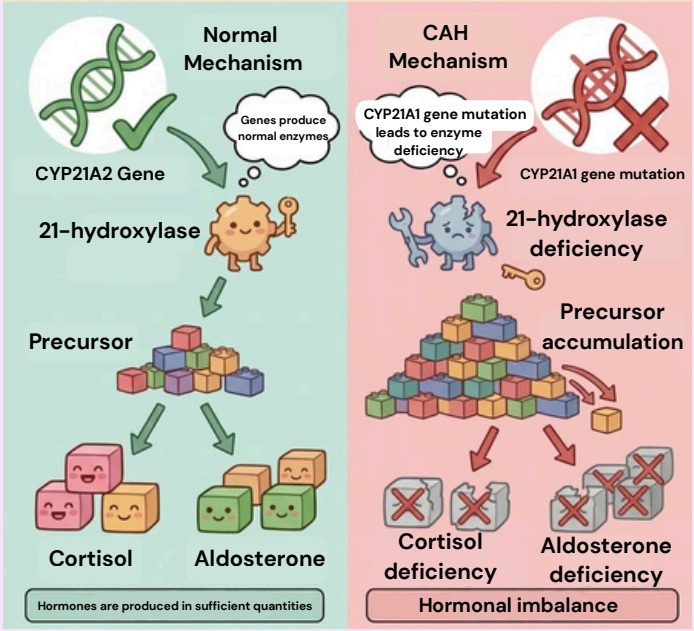
Hormones produced from adrenal glands affect body energy level, blood sugar levels, blood pressures, salt levels, body response to illness or stress, normal growth and genital development in boys and girls.



What is congenital adrenal hyperplasia?

Congenital Adrenal Hyperplasia (CAH) is an autosomal recessive disorder, with 90-95% of cases caused by CYP21A2 mutations resulting in 21-hydroxylase deficiency. This enzyme is essential for cortisol synthesis; its absence causes cortisol deficiency, hormonal imbalance, and precursor accumulation. Some patients also lack mineralocorticoids, leading to severe salt-wasting.

CAH Mechanism Diagram



Limitation of CAH screening

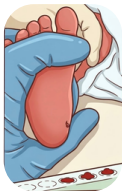
CAH screening test targets to screen out patient with severe phenotypes,(e.g. salt losing subtype of CAH). Patients with milder form of CAH presentation may escape from screening test and so they are not the target of this screening test.

What are the symptoms of CAH?

Low levels of cortisol and mineralocorticoids cause nausea, fatigue, dehydration, and weight loss. About 75% of CAH patients have the severe 'salt-wasting' form, placing infants at risk of salt-wasting crises, severe hypotension, shock, or death—especially during acute stress. Additionally, cortisol deficiency prompts the brain to overstimulate the adrenal glands, causing adrenal hyperplasia and excess androgen production, which can lead to atypical genital development in female infants.

	Salt-wasting type	Non-salt-wasting type
Hormonal Status:	Severe deficiency of both cortisol and aldosterone	Cortisol deficiency, aldosterone levels relatively preserved, with marked conversion of precursors into androgens
Impact:	Severe electrolyte imbalance (hyponatremia, hyperkalemia)	Electrolyte levels are normal
Manifestations:	Shock, frequent nausea and vomiting, severe hemodynamic instability	Precocious puberty and advanced bone age in both sexes, along with atypical external genital development in female infants

What is newborn CAH screening and its aim?



We aim to detect babies at risk of having CAH and institute early treatment before they develop potentially fatal salt-losing crisis. A few drops of blood are collected onto a card by pricking the baby's heel between 24 hours and 7 days of life.

The screening test detects a hormone precursor called 17-hydroxyprogesterone (17OHP) which is elevated in CAH patients but also in some other conditions.17OHP will drop in normal baby with time. The best time for collection is after 48 hours of life.

All babies with a positive screening result have to undergo further investigation to confirm whether they are affected by CAH or not.

What is the outcome of CAH patient?

Babies with CAH are treated by giving oral medications which replace the hormones that are missing in their bodies. With early and proper treatment, the life expectancy of CAH patients could be normal.

Early treatment can also reduce the chance of early puberty and short stature caused by excessive production of male sex hormone.

Affected girls with abnormal genital development may require corrective surgery.