Newborn Metabolic Screening Program
CUHK – BCM Joint Centre for Medical Genetics
The Chinese University of Hong Kong

新生兒代謝病篩查
香港中文大學 – 貝勒醫學院聯合醫學遺傳中心

Information For Parent

Screening for Congenital Adrenal Hyperplasia

Enquiries:
(852) 5569 6412 (office hour from 9-17:00)
(852) 3505 4219 (voice mail service available during non-office hours)
Website: http://www.obg.cuhk.edu.hk/fetal-medicine/fetal-medicine_services/iem/
If you wish to join this screening program, please contact your obstetrician during antenatal period or contact your paediatrician within 7 days after delivery.

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What are the functions of adrenal glands?

Adrenal glands are a pair of small organs located above the kidneys, which produce a right 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 a group of genetic disorders which the body cannot produce enough cortisol. Some patients may also have a lack of mineralocorticoids. The condition results in excessive production of male sex hormone (androgen). 90 – 95% of CAH are caused by deficiency of the enzyme 21-hydroxylase.

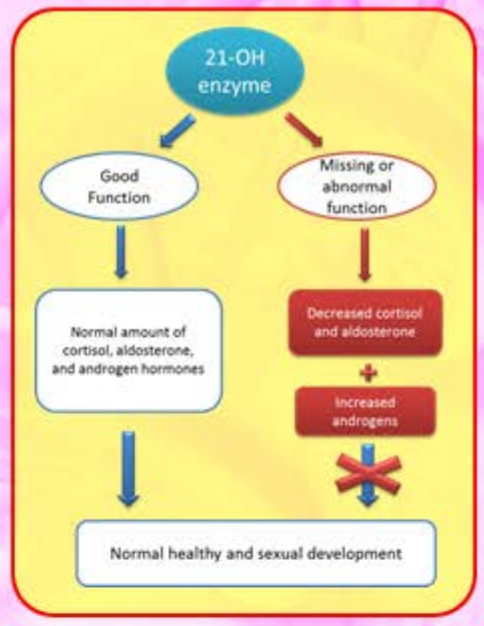
What are the symptoms of CAH?

Low level of cortisol and mineralocorticoids may cause nausea and vomiting, tiredness, dehydration, and weight loss. In the most severe case, CAH can lead to low blood pressure, shock or even death during illness or stress. These symptoms known as "salt-losing crisis" are seen in approximately 75% of CAH patients.

Excessive male sex hormone may cause abnormal genital development in girls.

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 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.