Newborn Metabolic and Congenital Adrenal Hyperplasia Screening Programs

新生嬰兒代謝及先天性腎上腺皮質增生症篩查計劃

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There is no fear in love. Instead, perfect love drives fear away
1 John 4:18a

Hong Kong Baptist Hospital is in collaboration with The Chinese University of Hong Kong (CUHK) for Inborn Errors of Metabolic (IEM) and newborn Congenital Adrenal Hyperplasia (CAH) Screening. The goal is to help affected babies by early diagnosis and treatment as well as to enhance the public awareness of the disease.

Introduction

A minority of newborns (1 in 4,000) may suffer from IEM, which if left undetected and untreated, could significantly affect the long-term health and development of the child.

What are Inborn Errors of Metabolism (IEM)?

IEMs are genetic defects which prevent some essential enzymes in the body from being produced. When the body cannot successfully break down (metabolize) some substances in food/milk such as fats, protein or sugar, there is deficiency of certain essential components required for growth and development, with accumulation of toxic metabolites in the body.

If these disorders are not treated early, they can lead to serious outcomes including learning difficulties, mental retardation, ill health and even death. These adverse outcomes are however potentially preventable nowadays, with effective screening test performed as early as few days after birth.

Who needs to be screened?

Every newborn baby is recommended to be screened unless the baby's health condition is not suitable.

What kinds of IEM are screened?

The test screens for 30 IEMs in the following categories:

- Amino acid disorders
- Fatty acid oxidation disorders
- Organic acid disorders

For more details on these 30 IEMs, please visit:

Please note that not all IEMs can be screened.

How to screen for and diagnose IEM?

A few drops of blood are collected onto a card by pricking the baby's heel after completing oral feeding for 1 day, and up to the 7th day of life. Results will be available within a few days.

How are results reported?

Results from the screening will be reported as follows:

Normal

This indicates that the baby has a very low chance of having one of the 30 IEMs. The majority of babies (98-99%) will have a 'Normal' result.

Positive

This indicates that the baby is at risk of having an IEM. Immediate referral to a Paediatrician for clinical evaluation, further diagnostic workup and management are needed.

Uncertain

This indicates that the sample analysis is inconclusive and a new sample is needed. This may happen in about 1% of all screened babies. Parents of babies with an 'Uncertain' result will be contacted to arrange for taking another sample within 14 days of life.

How accurate is the screening?

The accuracy for the screened IEMs is high. Similar to any laboratory screening test, there is a small chance that some affected infants may be missed (false negatives), while some unaffected infants may be wrongly identified (false positives). Therefore, it is extremely important that all abnormal screening results should be followed by standard diagnostic tests for confirmation.

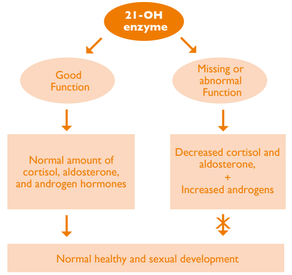
What are the functions of adrenal glands?

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

Hormones producing 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 are 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 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.

The aim of newborn CAH screening

To detect babies at risk of having CAH and institute early treatment before they develop potentially fatal salt-losing crisis.

What is newborn CAH screening?

The screening test detects a hormone precursor called 17-hydroxyprogestrone (17OHP) which is elevated in CAH patients but also in some other conditions. 170HP will drop in normal baby with time.

A few drops of blood are collected onto a card by pricking the baby's heel between 24 hours and 7 days of life. Results will be available within a few days.

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.

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