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

Introduction

Every parent anticipates the birth of a healthy baby. However, a minority of newborns (1 in 4000) may suffer from '**Inborn Errors of Metabolism**' (IEM), which if left undetected and untreated, could significantly affect the long term health and development of the child. To this end, The Chinese University of Hong Kong (CUHK) now offers an expanded newborn screening program which can provide early detection of an extensive range of metabolic diseases previously untested for in Hong Kong.

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 is Inborn Errors of Metabolism?

IEMs are genetic defects which prevent some essential enzymes in the body from being produced. As a result, there is deficiency of certain essential components, or accumulation of toxic substances in the body. If these disorders are untreated, they can lead to serious outcomes including learning difficulties, mental retardation, organ dysfunction and even death. Some of these conditions are however potentially treatable 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 31 IEMs belonging to the following categories:

For more details on these 31 IEMs, please visit the website. *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 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.

How are results reported?

Results from the screening will be reported as either:

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

2) **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.

3) **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 another sample to be taken.

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