## Your baby's result is Positive for one of the conditions.

Having a positive result does not necessarily mean that your baby has a disorder. It only means that further testing is needed. You will be contacted to make arrangements for a metabolic physician to review and investigate. If a diagnosis is made, your baby will be given appropriate treatment. It is important to know that most babies in Singapore are screened negative. Rare but life-threatening complications of the disorders can usually be prevented with early diagnosis and treatment.

### Get in Touch

For more information about the metabolic screen in Singapore, please ask your paediatrician.

Alternatively, you can contact us at:

### A/Prof Denise Goh

denise\_li\_meng\_goh@nuhs.edu.sg

### Dr Chin Hui-Lin

hui-lin chin@nuhs.edu.sg

Your baby will be screened as a part of routine newborn care. Please note that you have the right to refuse the metabolic screen. If this is your wish, please speak to your healthcare provider.

Content in this brochure is used with written permission from KK Women's and Children's Hospital.

The information provided in this publication is meant purely for educational purposes and may not be used as a substitute for medical diagnosis or treatment. You should seek the advice of your doctor or a qualified healthcare provider before starting any treatment or if you have any questions related to your health, physical fitness or medical condition.

Information is correct at the time of printing (May 2020) and subject to revision without prior notice.

# About the Khoo Teck Puat - National University Children's Medical Institute (KTP-NUCMI)

The KTP-NUCMI is the paediatric arm of the National University Hospital and comprises the Departments of Paediatrics, Paediatric Surgery and Neonatology. We provide comprehensive and specialised medical and surgical services for newborns, children and adolescents. NUH is the only hospital in Singapore that offers paediatric kidney and liver transplant programmes. Through a generous gift from the Estate of Khoo Teck Puat, we have set up an integrated outpatient facility with medical, diagnostic and rehabilitation services.

For more information about us, visit www.nuh.com.sg/ktp-nucmi.

### Contact Us

### 24-hour Children's Emergency

Location: NUH Main Building, Zone F, Level 1 General Enquiry: +65 6772 5000

#### KTP-NUCMI

Location: NUH Main Building, Zone E, Level 2

(Accessible via Kent Ridge Wing, Zone C, Level 2, Lift Lobby C)

Operating Hours: 8.30am - 5.30pm (Mon to Fri), 8.30am - 12.30pm (Sat)

General Enquiry: +65 6772 5736 Appointment Line: +65 6908 2222

Email: ktpnucmi\_appt@nuhs.edu.sg

### 9a Viva-University Children's Cancer Centre

Location: NUH Medical Centre, Zone B, Level 9 Operating Hours: 8am – 5.30pm (Mon to Fri)

Appointment Line: +65 6772 5030 Fax: +65 6872 4314

Email: cbccappt@nuhs.edu.sg

#### NUH Children's Urgent Care Clinic @ Bukit Panjang

Location: Junction 10, 1 Woodlands Road, #01-22, Singapore 677899 Operating Hours: 9am - 11pm daily (including public holidays) General Enquiry: +65 6219 1538 Email: childrenucc@nuhs.edu.sg Website: www.nuh.com.sg/ucc

### NUH Child Development Unit @ JMC

Location: Jurong Medical Centre,

60 Jurong West Central 3, Level 2, Singapore 648346

Operating Hours: 8.30am - 5.30pm (Mon to Fri)

Appointment Line: +65 6665 2530/2531 Fax: +65 6665 0158

Email: cdu@nuhs.edu.sg Website: www.nuh.com.sg/cdu

### NUH Child Development Unit @ Keat Hong

Location: Keat Hong Community Club,

2 Choa Chu Kang Loop, #03-01, Singapore 689687

Operating Hours: 8.30am - 5.30pm (Mon to Fri)

Appointment Line: +65 6769 4537/4637 Fax: +65 6665 0158

Email: cdu@nuhs.edu.sg Website: www.nuh.com.sg/cdu

### National University Hospital

Location: 5 Lower Kent Ridge Road, Singapore 119074

Tel: +65 6908 2222 Email: contactus@nuhs.edu.sg

Website: www.nuh.com.sg



Scan QR code for NUH Campus Map

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# **Screening Tests For Your Newborn Baby**



Khoo Teck Puat – National University
Children's Medical Institute

# A New Beginning of Life

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Congratulations on the arrival of your new bundle of joy. As a parent, your baby's health is your first priority. To ensure that your baby gets the best start in life and stays healthy, your newborn can be screened for more than 40 rare medical disorders.



Although most newborns with these disorders look healthy at birth, they may be at risk of having serious health problems later in life. These may include learning difficulties, recurrent sickness and even death if their disorder is not detected and treated early.

## Early Detection Allows Early Treatment

These disorders are rare and they affect about one in 3000 births in Singapore. By testing newborns during their first few days of life, such disorders can be treated early, often before any sign or symptom surfaces. This pamphlet will provide you with information about newborn screening in Singapore, its benefits to your newborn, and answer some questions you may have in mind.

The disorders in the newborn screening include:

### Inborn Errors of Metabolism

When the body cannot successfully breakdown (metabolise) some substances in food/milk such as fats, protein or sugar, they can accumulate and become toxic to the body. This can cause serious health problems.

The newborn screening looks for more than 30 metabolic related disorders in the following categories:

- o Organic Acid Disorders
- Fatty Acid Oxidation Disorders
- Amino Acid Disorders

### Cystic Fibrosis

This is a genetic condition that leads to accumulation of thick mucus in different organs leading to severe chest infections and poor growth.

### • Congenital Adrenal Hyperplasia

This is a disorder of hormone production that may affect baby's metabolism, response to infection, ability to regulate salt levels and sex characteristics.

### Severe Combined Immune Deficiency (SCID)

This condition results in extremely poor immunity and the child can develop severe infections.

### Galactosemia

Babies with galactosemia are unable to process lactose containing milk feeds and ingestion of such milk may lead to liver failure.

### • Biotinidase Deficiency

This is a metabolic disorder whereby the body is unable to recycle biotin leading to a deficiency. This can result in complications such as seizures, developmental delay and low muscle tone. Babies can also have skin and hair abnormalities

IT IS IMPORTANT TO NOTE THAT ALTHOUGH THESE ARE ALL SERIOUS MEDICAL CONDITIONS, EARLY DIAGNOSIS AND TREATMENT CAN LEAD TO A BETTER OUTCOME.

### A Small Test with Big Benefits

In order to perform the screening tests, a small blood sample is needed from your baby. A few drops of blood are collected on special filter paper by pricking the heel. The sample is sent to the National Expanded Newborn Screening Laboratory for analysis. Blood samples can be taken from your baby any time between one day (24 hours) and seven days old. The best time for collection is between one to three days old (24 to 72 hours).

### High Risk or Low Risk Results

When the sample has been analysed and results are available, one of the following will occur:

## Your baby's result is Negative for all the disorders.

More than 99 per cent of the babies fall into this category. It means that your baby is at very low risk of having these disorders. The result will be filed into your baby's medical record. On very rare occasions, a disorder may not be picked up on the metabolic screen.

### • A repeat sample is required.

A second sample is needed for about one in 1000 babies. It is usually needed because the first sample showed borderline results, which means it was not possible to tell whether or not a problem exists. You will be contacted to arrange for another sample to be taken.