Newborn
SCID
Screening

# Newborn Screening Programme

*for*

# Severe Combined Immune Deficiency

# (SCID)

醫院管理局
HOSPITAL AUTHORITY

# Newborn Screening for SCID at a Glance

1 **Antenatal Visit**

Antenatal health education related to Newborn Screening Programme for SCID is provided by healthcare professionals.

2 **Consent**

With parent's written consent after delivery, majority of babies will receive heel pricking procedure for blood specimen collection at 24-72 hours after birth.

3

Collection of blood specimen on a filter paper card.

4 **Laboratory**

Laboratory analysis

5 **Screening results**

Screening results of SCID available.

6 **Report**

Results will usually be available within 7 working days.

Normal results: Parents will not receive notification.

For abnormal or uncertain results: Parents will be informed early by phone. Babies will be referred to paediatricians for further evaluation.

## What is Newborn Screening?

Through the provision of screening test to newborn babies, it is intended to achieve early diagnosis of serious yet treatable disorders which may not have obvious symptoms at the early stage, so as to reduce morbidity and mortality.

The Pilot Newborn Screening (NBS) for Inborn Errors of Metabolism (IEM) was first introduced in 2015 in Hong Kong through a pilot scheme launched with the Hospital Authority. Nowadays, NBS for IEM and Severe Combined Immune Deficiency (SCID) have become regular services provided to all babies born at the eight Public Hospitals with Obstetrics service.

Starting from October 2023, the Hospital Authority will launch the Pilot NBS for SMA.

## What is SCID?

The immune system protects us from pathogens like bacteria, viruses, fungi and protozoa. The immune system consists of white blood cells such as lymphocytes which develop and mature in the bone marrow and various lymphoid organs such as the thymus, tonsils, lymph nodes and spleen. Defective immune response leads to susceptibility to infections, immune dysregulation, inflammation or even cancers.

Primary immune deficiencies (PIDs) are a group of inborn errors of immunity caused by hereditary genetic defects of the immune system. To date, more than 400 PIDs are known. SCID is considered to be the most serious form of PIDs. It is characterised by a combined absence of T-cell and B-cell functions, leading to very low immunity. Infants born with SCID can become sick very quickly from common illnesses like a cold or stomach virus, and they are at risk of developing serious infections such as pneumonia, enteritis, candidiasis or even systemic dissemination of bacteria and fungi. Fortunately, effective treatments such as bone marrow transplantation and gene therapy can restore the faulty immune system and cure this otherwise fatal disease.

## Why is SCID Newborn Screening Important?

SCID is a rare disorder. The estimated incidence is 1 in 50,000 newborns in Hong Kong. Most babies with SCID appear healthy at birth, and very often nothing abnormal is detected at antenatal check-ups. Although SCID is a genetic disorder, a positive family history is not always present. Therefore, even if the parents and family members are all healthy, SCID can occur in any newborn.

Owing to the lack of specific signs and symptoms at the onset of infections, SCID is not usually suspected by medical professionals. Once infection sets in, it is extremely difficult to clear despite anti-microbial therapy, and may cause organ damage. Affected infants may run into life-threatening complications, often beyond salvage. On the contrary, the chance of survival and cure is excellent if SCID could be diagnosed before the onset of infection. The latest medical technologies make early detection possible so that affected babies can be diagnosed in the asymptomatic stage and be given the necessary life-saving therapies.

## What is the Scope of this Screening Programme?

The NBS for SCID is coupled with the NBS for IEM and SMA offered to all newborn babies delivered in public hospitals. The small amount of blood specimen obtained by heel prick will be sufficient for all NBS tests for the above-mentioned conditions.

SCID newborn screening makes use of the detection of T-cell receptor excision circle (TREC), which is a molecular marker for naïve T-cells. Hence, the quantification of TREC from heel prick can be regarded as a surrogate marker for newly produced T-cells. If TREC is below the normal range, this would suggest reduced T-cell number and confirmation by further blood tests is required to exclude SCID or other disorders of T-cell lymphopenia. For the latter, the immune defects are often not as severe as SCID, but are important to investigate, monitor, and treat as appropriate.

## Is My Baby Eligible for this Screening Programme?

All babies born at the eight Public Hospitals with Obstetrics service are eligible for the screening programme, as long as a written consent is signed by a parent. Participation is voluntary and free of charge.

## What is the Screening Process like?

The screening process for SCID is conducted along with IEM and SMA, including health education, blood sample collection, laboratory testing, confirmation of screen-positive (abnormal or uncertain) cases and referral for follow-up care. Newborn babies with positive screening result will be referred to the Hong Kong Children's Hospital for evaluation and follow-up by specialists in paediatric immunology. If you consent to newborn screening for IEM, SCID, and/or SMA, blood specimen by heel pricking is collected only once and the screening tests for all the above-mentioned conditions will be performed on the same specimen. Newborn babies under the following conditions require additional blood specimens for testing*:

1. prematurity (less than 34 weeks of gestation), or
2. birth weight less than 2kg, or
3. being admitted into Neonatal Intensive Care Unit (NICU).

**All blood samples are to be sent to the laboratory under Hospital Authority for testing.*

## *Screening results and follow-up*

| Screening Results | | Follow-up Action |
|---|---|---|
| **Normal** | **Risk** of suffering from SCID is very **low**. | Parents will not receive any notification. |
| **Abnormal** | **Risk** of suffering from SCID is **high**. | Hospital staff will notify parents by telephone **within 7 working days**. Blood tests will be arranged for further diagnostic testing. |
| **Uncertain** | Less than 1% of the specimen. | |

## *Diagnostic testing*

For **abnormal** or **uncertain** screening results, follow-up action for confirming T-lymphocyte number is required. If this happens, you will receive a phone call informing you to bring your baby to the Hong Kong Children's Hospital for evaluation by specialists, followed by blood taking for enumeration of T-cells. Subsequent services will be charged as admission or attendance at the specialist outpatient clinic under the Hospital Authority accordingly.

If T-cell number is abnormal, further investigations will be arranged to find out the cause and follow-up by specialists in paediatric immunology will be provided. If T-cell number is normal, no further follow-up will be needed.

## *Treatment arrangement*

Babies who are confirmed having SCID will be admitted to hospital for anti-microbial therapy, immunoglobulin replacement therapy and preparations for haematopoietic stem cell transplantation (HSCT).

## How Accurate is the Screening Test?

NBS for SCID has a very high accuracy rate. However, as with any screening test, NBS may miss a baby with the disease. Some babies with SCID may have a negative screening result (i.e. false negative). Hence, a normal screening result only suggests the chance of having SCID is extremely low, but cannot be taken that the baby is normal in all aspects. If a baby has symptoms of SCID, diagnostic testing for the disease is still recommended even if the baby had a negative result for SCID.

On some occasions, a NBS screening test is positive for a disease but follow-up testing shows the baby does not have the disease (i.e. false positive). Thus, an abnormal or uncertain NBS screening result just means that the baby needs further evaluation by a paediatrician, and not necessarily implies that the baby has the disease.

## Enquiries

For general queries, please call:

5741 4280 (Clinical Genetics Service Unit, Hospital Authority)

For further enquiries about this NBS for SCID, please approach your healthcare professionals when attending antenatal visits.