Pilot Newborn Screening Programme

for

Spinal Muscular Atrophy (SMA)

醫院管理局
HOSPITAL AUTHORITY

Newborn Screening for SMA at a Glance

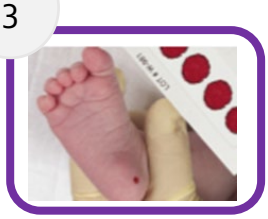
1 **Antenatal Visit**

Antenatal health education related to Pilot Newborn Screening Programme for SMA 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 SMA available.

6 **Report**

Results will usually be available within 7 working days.

Normal results: Parents will not receive any 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 tests 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 SMA ?

SMA is a hereditary neuromuscular disorder due to mutations or defects in the survival motor neuron 1 (SMN1 gene) causing progressive degeneration and cell death of the motor neurons in the spinal cord and lower brainstem. When there is a disruption in the signals between motor neurons and the skeletal muscles, the muscles become weakened and atrophic. The affected patients will have progressive difficulty in the control of movement, eating, and breathing. SMA presents in infancy in the most severe form with the symptom onset before age of 6 months. They have difficulty sitting, breathing, and eating. Without gastrostomy for feeding support and ventilation for breathing support, most children die before the age of 2 years.

For early childhood SMA, symptoms onset is usually between age of 7 to 18 months. The affected patients cannot walk and gradually lose the ability to sit. Most of them develop severe scoliosis. They also have difficulties in breathing and swallowing.

Late childhood and adolescence SMA have their symptom onset after age of 1.5 years. They can walk but over time they have increased difficulty in walking. Some of them eventually lose their ability to walk and develop scoliosis, but their life expectancy will not be shortened.

Adult-onset SMA is rare, and the disease presentation and progression are usually mild.

Fortunately, SMA disease-modifying treatments are now available, and the earlier the treatment the better the treatment outcome.

Why is SMA Newborn Screening Important?

Owing to the lack of specific signs and symptoms at the onset of the disease, SMA is not usually suspected by parents or even medical professionals at the early stage. SMA can deteriorate quickly, and the degenerated motor neurons are hard to recover. Early treatment at the pre-symptomatic stage can preserve the survival of more motor neurons and so the motor ability hence can have the best clinical outcome.

The latest medical technologies make early detection possible so that affected babies can be diagnosed in the pre-asymptomatic stage and be given the necessary early SMA disease-modifying therapy. Early treatment not only is life-saving, but also can prevent the development of severe complications from SMA, and lead to much better motor performance and a healthy life for the affected children.

What is the Scope of this Pilot Screening Programme?

The pilot NBS for SMA is coupled with the existing regularised NBS for IEM and SCID 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.

SMA newborn screening makes use of the detection of mutations or defects in both copies of theSMN1 gene.

Is My Baby Eligible for this Pilot 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 SMA is conducted along with that for IEM and SCID, 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 neurology. If you consent to newborn screening for IEM, SCID and 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 the Hospital Authority for testing.*

Screening results and follow-up

Screening Results		Follow-up Action
Normal	**Risk** of suffering from SMA is **very low**.	Parents will not receive any notification.
Abnormal	**Risk** of suffering from SMA 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, you will receive a phone call informing you to bring your baby to the Hong Kong Children's Hospital for evaluation by specialists. Subsequent services will be charged as admission or attendance at the specialist outpatient clinic under the Hospital Authority accordingly.

Treatment arrangement

Babies who are confirmed having SMA will be admitted to the hospital or consulted at the specialist outpatient clinic for treatment. Subsequent services will be charged as admission or attendance at the specialist outpatient clinic under the Hospital Authority accordingly.

How Accurate is the Screening Test?

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

On some occasions, an 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 Pilot NBS for SMA, please approach your healthcare professionals when attending antenatal visits.

XXX 2023