Joshua Hellmann Foundation

Newborn Metabolic Screening Program

CUHK-BCM Joint Centre for Medical Genetics

The Chinese University of Hong Kong

Information for Parents

Screening for

Spinal Muscular Atrophy

(SMA)

Enquiries:

(852) 5569 6412 (office hours from 9:00-17:00)

(852) 3505 4219 (voicemail service during non-office hours)

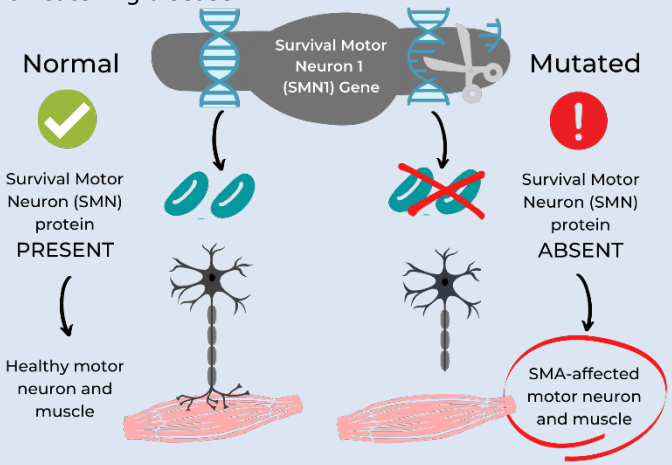
Website: http://www.obg.cuhk.edu.hk/fetal-medicine/fetal-medicine_services/iem/

If you wish to join this screening program, please contact your obstetrician during antenatal period or contact your paediatrician within 7 days after delivery.

What is spinal muscular atrophy?

Spinal muscular atrophy (SMA) is a rare genetic disorder. The most common form of SMA is caused by mutations i.e. changes in the survival motor neuron 1 gene (SMN1). SMN1 is essential for producing the survival motor neuron (SMN) protein, which is needed for the survival and proper functioning of motor neurons. For patients with SMA, the production of SMN protein is impaired.

Without a sufficient level of SMN protein, motor neurons break down and die. As a result, signals cannot be transmitted between the brain and muscles, leading to muscle weakness and gradual wasting of the skeletal muscles. SMA may sometimes affect muscles involved in feeding, swallowing and breathing making it a life-threatening disease.



What are the symptoms of SMA?

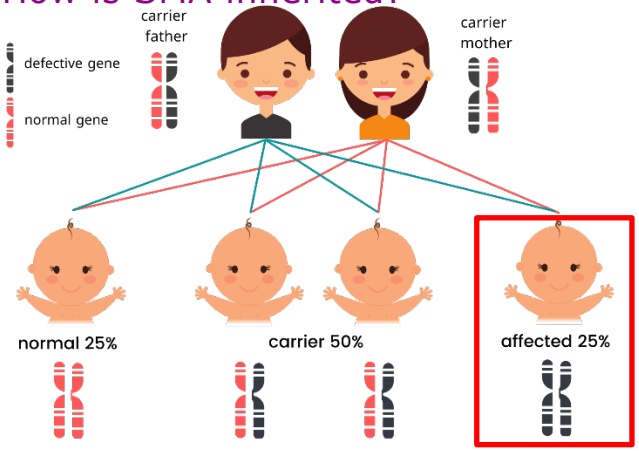
SMA is classified into 4 types based on the age of onset, symptoms and rate of progression. Typical symptoms of SMA include floppiness, movement difficulties, tremors, swallowing and breathing problems, etc.

Type 1 is the most severe form of SMA. Patients present at birth or within the first 6 months of infancy. Infants with Type 1 SMA fail to sit up, have a weak cry, and have difficulties in breathing, feeding and swallowing.

Type 2 SMA is less severe than Type 1. Patients usually present between 7 and 18 months old. These infants can sit on their own but may fail to walk independently.

Types 3 and 4 SMA are relatively milder forms. Patients present after 18 months old and are capable of walking properly.

How is SMA inherited?



SMA is an autosomal recessive disease, which means a baby needs to inherit one defective gene for SMA from each of its parents to develop the condition. If both parents are carriers of the defective gene, each of their baby will have a 25% chance of having SMA, 50% chance of being a carrier without any symptoms, and 25% chance of not carrying a defective gene at all.

SMA screening and its aim

The SMA screening aims to detect babies at risk of SMA as early treatment can prevent severe disability. A few drops of blood, from which SMN1 copy number is measured, are collected onto a card by pricking the baby's heel in its first 24 hours to 7 days of life. All babies with a positive screening result will have to undergo further investigations, e.g. SMN2 copy number to determine the severity of the codition and which type of SMA develops.

Outcomes of SMA patients

There is currently no complete cure for SMA. However, early diagnosis can help relief the progression of the disorder. Physiotherapy, occupational therapy and rehabilitation may help improve the posture of patients, prevent joint immobility, and slow muscle weakness. Proper diet is essential to maintaining weight and strength whilst prolonged fasting should be avoided. Nusinersen is an approved and registered drug in Hong Kong that helps increase the production of SMN protein in SMA Type 1 infants and Type 2 children with a late onset.