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

Severe Combined Immunodeficiency (SCID)

Enquiries:

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

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

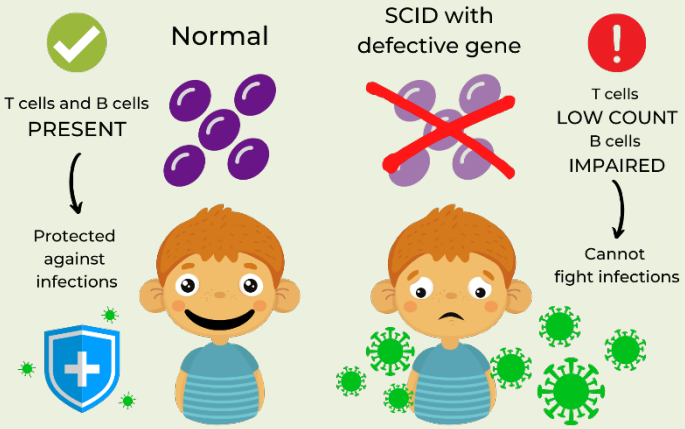
Website: http://www.obg.cuhk.edu.hk/fetal-medicine/fetal-medicine_services/iem/

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 severe combined immunodeficiency?

Severe combined immunodeficiency (SCID) is a group of rare genetic disorders characterised by a profound impairment in the immune system involving two types of white blood cells: T cells which directly attack viruses; and B cells which produce antibodies to recognise the invasion of microbes and destroy them with the aid of T cells.

White blood cells fail to develop properly in SCID patients carrying various defective genes. Therefore, with a severe defect in T-cell development and malfunction of B cells, the patient's immune system has trouble defending the body against bacteria, viruses and fungi, leading to recurrent infections which can be life-threatening.



What are the symptoms of SCID?

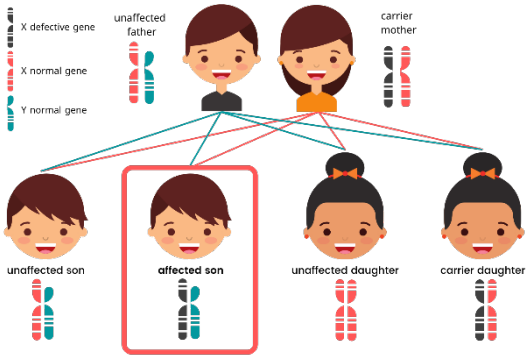
Babies with SCID are asymptomatic at birth as they receive antibodies from their mothers during pregnancy. However, they gradually develop symptoms between 2 to 6 months of infancy as the level of maternal antibodies drops.

Symptoms of SCID may vary among patients. Common symptoms include chronic diarrhoea, recurrent infections, poor growth. Organisms that usually cause mild or no illnesses in normal people may cause life-threatening infections in SCID patients.

How is SCID inherited?

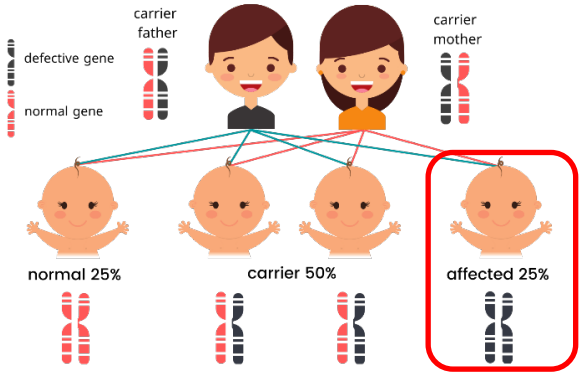
There are two modes of inheritance for SCID. The more common form is X-linked recessive inheritance. A male infant carrying such mutated gene is affected as he carries only one X chromosome.

X-linked Recessive Inheritance



The other mode of inheritance is autosomal recessive inheritance which means an infant must inherit one defective gene of SCID from each of its parents to develop the condition. If both parents are carriers, each of their child will have a 25% chance of having SCID regardless of gender.

Autosomal Recessive Inheritance



SCID screening and its aim

The SCID screening aims to detect babies at risk of SCID as early detection allows for proper isolation from infection and referral for treatment. A few drops of blood are collected onto a card by pricking the baby's heel in its first 24 hours to 7 days of life, and the level of T-cell receptor excision circles (TREC) is measured. TREC are produced when our body produces new T cells. As T cell level is low in babies with SCID, the level of TREC would also be low in them. All babies with a positive screening result will have to undergo further investigations to confirm whether they are affected by SCID or not.

Outcomes of SCID patients

A better outcome can be achieved if bone marrow transplant from a healthy and matched donor can be performed at an early age. While waiting for the bone marrow transplant, it is critical to employ protective measures to prevent the patient from contracting infections. They may also need regular antibody infusions under doctor's supervision.