Alberta Health Services

Newborn Metabolic Screening

*Information for Health Professionals*

# Severe Combined Immunodeficiency (SCID)

## What is SCID?

Severe combined immunodeficiency (SCID) is a group of genetic disorders that cause severe abnormalities of the immune system. Infants with SCID are highly susceptible to developing recurrent and life-threatening infections.

## What causes SCID?

SCID is a severe immune defect caused by a variety of genetic variants that impair the development and/or function of T, B and/or NK cells. T and NK cells are important in fighting viral and fungal infections, and B cells produce antibodies, which primarily protect against bacterial infections. Consequently, the ability of the immune system to fight infection is reduced or absent.

## How common is SCID?

SCID is a rare condition with an estimated incidence of 1 in 70 000 infants born in Canada. SCID is more common in First Nations, Metis and Inuit infants, with an estimated incidence of 1 in 20 000 births.

## What are the clinical features of SCID?

Infants with SCID usually appear normal at birth. Diarrhea, failure to thrive, recurrent serious and/or opportunistic infections commonly occur in the first six months of life.

## What is the screening test for SCID?

Screening for SCID is performed by measuring blood spot T cell receptor excision circles (TRECs), byproducts of T cell development. Newborn screening can detect some other types of immune deficiencies, such as DiGeorge syndrome or ataxia telangiectasia. Newborn screening will not detect all infants with SCID. Infants with symptoms or signs consistent with SCID require timely assessment and diagnostic testing even if their newborn screen result is normal for SCID.

## How is the diagnosis of SCID confirmed?

The diagnosis of SCID is confirmed by measuring lymphocyte counts to assess the presence/absence of T and B lymphocytes in blood. Other immune testing and molecular genetic analysis of SCID genes may be performed. Specialists at the clinics listed below will arrange diagnostic testing.

## How is SCID treated?

SCID is treated with immunoglobulin infusions, prophylactic antibiotics and isolation precautions to protect infants from infection. Breastfeeding is interrupted as there is a risk of infection transmitted from the mother. Bone marrow transplantation or other therapies may also be considered.

## What is the outcome of treatment for SCID?

Early treatment can reduce the number of serious infections and improve life expectancy. Prompt curative therapies, such as bone marrow transplantation, are associated with excellent prognosis and long-term quality of life. The clinical outcome for untreated patients is very poor.

## Is SCID inherited?

SCID is a genetic condition that is most commonly inherited as an autosomal recessive disorder or as an X-linked disorder. Parents of a child with SCID may be carriers and may be at increased risk of having another affected child in subsequent pregnancies. Genetic counselling and prenatal testing are available to all families with SCID.

For additional resources, please call:

### Immunology Clinic (Edmonton)

Stollery Children's Hospital
8440 – 12 Street
Edmonton, AB T6G 2B7
Tel: 780-407-7909
Fax: 1-888-775-9068

Emergency consultations:
Phone 780-407-8822 and ask for the pediatric immunologist on call.

### Hematology/Immunology Clinic (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Tel: 403-955-7155
Fax: 403-955-7393

Emergency Consultations:
Phone 403-955-7211 and ask for the immunologist on call.