Quick Guide

Severe Combined Immune Deficiency (SCID) Screening

Background

Severe Combined Immunodeficiency (SCID) is a life-threatening genetic condition in which affected babies are unable to fight even simple infections. SCID is often referred to as the 'bubble boy disease' highlighting the threat of exposure to germs and is considered an immunological emergency. Without appropriate timely treatment, most die before the age of 2 years. If diagnosed early and then treated early, SCID can be entirely curable. Early diagnosis is possible with universal SCID newborn screening.

Babies with SCID initially look perfectly healthy until they suddenly become sick, often around the age of two to three months. As SCID is a rare condition (one in about 50,000), the diagnosis may not be considered, and testing not undertaken before it is too late. Some babies do not survive this initial illness despite starting state of the art therapies.

SCID can be cured by replacing the faulty immune system with a healthy normal one by bone marrow transplantation. If performed before the baby gets sick, especially before the age of three and a half months, there is a greater than 90% survival. Transplantation performed later, especially having presented with infection reduces the chance of survival to about 70%.

International cost effectiveness studies have also demonstrated that the cost of treating and transplanting children presenting symptomatic from SCID far outweighs the cost of transplanting asymptomatic babies, supporting the economic argument for screening. These costs include those related to prolonged hospitalisations and long-term organ damage that may not be reversible.

So early diagnosis is essential to ensure the best health outcomes and quality of life for these babies. Just like other severe illnesses where children are born looking well but then develop devastating illness, there is a test that can be done on the newborn blood spot that is taken in all babies in the days after birth.

SCID fulfils the internationally recognised criteria for a clinical condition to be screened at birth through this process. SCID newborn screening is already standard practice in many countries including the USA, Canada, Norway, Spain, New Zealand, and as of mid 2024, all states of Australia.

Additional Information About SCID

Incidence

- The estimated **UK incidence rate** is **1:35,000 births** (Gaspar, 2011). This estimate is based the number of SCID cases presented to the two UK centres for care of SCID from 2008 and 2009 (20 per year). It therefore does not account for children who may have been diagnosed after death at other UK centres or children who died and were undiagnosed (Bazien Ltd, 2012).
- **Australian estimate of SCID 1:50,000 births**

Presentation

- Generally, between the ages of 3-6months the infant will present with symptoms such as persistent thrush, chronic diarrhoea, failure to thrive, eczematous rash, and recurrent severe and life-threatening infections.
- As SCID is a rare disease, the journey to diagnosis is often long and tumultuous with many other illnesses needing to be ruled out before SCID is considered and diagnosed. This is a stressful and traumatic period for the child and family.

When should it be detected?

- Between birth and 3 months, when the child is healthy

Why?

- So lifesaving Haematopoietic Stem Cell Transplantation can occur (HSCT) BEFORE the child shows symptoms. This is the optimum time and gives the optimum outcome of a cure.

How is it detected?

- Laboratory test: Levels of T and B cell numbers are detected using an assay for TREC (T-cell receptor excision circles) and KREC (Kappa recombination excision circles) which are small circles of DNA created during maturation of the cells.
- An absence of T cells indicates SCID.

Impact

- serious infections, extensive hospital stays, long term organ damage and eventually death.
- IDFA also recognises the impact this has on the family of the infant.

Current as of September 2025