PO Box 969
Penrith NSW 2751
E: info@idfa.org.au

IDFA
Immune Deficiencies Foundation
Australia

W: www.idfa.org.au
P: 1800 100 198
M: 0409 945 114

Immune Deficiencies Foundation Australia

Severe Combined Immune Deficiency (SCID) Newborn Screening Position Statement

Commonly known as the "bubble boy" disease, Severe Combined Immune Deficiency (SCID) is a **paediatric emergency**. It is the **most severe** form of inherited **primary immunodeficiency**, a life-threatening condition in which the body is unable to fight off serious infections.

This condition **is curable** with haematopoietic stem cell transplantation (HSCT) if treated in time, with best results **in the first months of life**.

For babies born with SCID, a delay in diagnosis may have fatal consequences. These babies initially look perfectly normal. Around the age of 2 to 3 months, they suddenly become extremely ill. As SCID is a rare condition, the diagnosis may not be considered and the special tests required for confirmation done before it is too late. The optimum time for these tests to be done is **before** the baby gets sick, preferably soon after birth. This enables commencement of early treatment to prevent the baby from deteriorating, increasing the chance of successful curative treatment.

The Immune Deficiencies Foundation of Australia recognises the severe impact undiagnosed and untreated SCID has on the infant, including 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.

Early identification, diagnosis and treatment of infants with disorders identified through **Newborn Screening** (NBS) **is vital** to preventing significant morbidity and mortality.

NBS for SCID prevents infant deaths from SCID and **increases the chance of successful cure** by Haematopoietic stem cell transplantation (HSCT).

SCID fulfils all the internationally recognised criteria for a clinical condition to be screened for at birth through NBS using the Standard Guthrie (dried blood spot) sample.

NBS for SCID should be mandatory in all States and Territories of Australia, as it is in New Zealand, Norway, Ontario, Canada, Spain, Qatar (private clinics), Singapore (private clinics) and most States of America. There are pilot studies in Japan, France, Italy, Germany, Saudi Arabia, Slovenia and Sweden.

IDFA supports the introduction of Newborn Screening for SCID in Australia to prevent further infant deaths and the enablement of curative treatment within the first months of life to ensure the best health outcome and quality of life of infant patients.

More information can be found at:
http://www.idfa.org.au/support-newborn-screening-for-severe-combined-immune-deficiency-scid/

"I wholeheartedly support the introduction of Newborn Screening for SCID in Australia. Newborn Screening for SCID may have saved my son's life. My hope is that with the introduction of this important test that no more babies will die from this condition"
Bianca (mother)

"If Australia had Newborn Screening for SCID, my baby's SCID would have been diagnosed earlier reducing the risk of multiple infections and his HSCT treatment would have been conducted earlier, improving his current quality of life"
Tracey (mother)