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Immune Deficiencies Foundation Australia (IDFA)

# Position Statement: Newborn Screening for Severe Combined Immune Deficiency

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 infection.

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

When a baby is born with SCID, a delay in diagnosis may have fatal consequences. These babies initially look perfectly healthy until they suddenly become sick, often around the age of two to three months. As SCID is a rare condition, the diagnosis may not be considered, and the required tests not undertaken before it is too late. Completing these tests before the baby becomes sick, preferably soon after birth, enables early diagnosis and commencement of early treatment to prevent the baby from deteriorating, increasing the chance of successful curative treatment.

The Immune Deficiencies Foundation of Australia (IDFA) recognises the severe impact undiagnosed and untreated SCID has on the infant, including serious infections, extensive hospital stays, long term organ damage and eventual death. IDFA also recognises the significant impact this has on the family of the infant.

**Newborn screening (NBS) in Australia** is vital in preventing significant child morbidity and mortality through the **early identification**, **diagnosis and treatment** of infants with disorders.

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

SCID fulfils 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, Qatar (private clinics), Canada, Spain and all but two States of America. There are pilot studies in Japan, France, Italy, Germany, Saudi Arabia, Slovenia and Sweden and the first Australian pilot study commenced in NSW in August 2018.

**IDFA supports the introduction of NBS 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 outcomes and quality of life for these infant patients.**

*For more information visit the IDFA website - www.idfa.org.au/what-is-pid/*

**Supported by:**

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*"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 of Harrison who passed away from SCID aged 5 mths).*

*"If Australia had NBS 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 of Harrison, SCID survivor).*