

EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Severe Combined Immunodeficiency (SCID) to the Recommended Uniform Screening Panel (RUSP) in 2010.

About the condition

SCID is the name of a group of serious disorders affecting the immune system. About 1–3 out of every 100,000 babies has SCID. Children with SCID are born with changes in important immune system genes. Because of these changes, children with SCID are missing key cells in the immune system. These changes weaken the immune system and make children with SCID prone to infections. Babies with SCID appear normal. There are many types of SCID, and each can cause problems with the immune system leading to serious infections. This can lead to death within the first year.

Treatment for SCID

Early diagnosis allows early treatment and improves outcomes for babies with SCID. Treatments that help to restore the immune system include hematopoietic stem cell transplantation and enzyme replacement therapy. The treatment a baby receives depends on many things, like the baby's type of SCID.

Detecting SCID in newborns

Newborn screening for SCID can happen along with routine newborn screening for other conditions during the first few days of life. SCID screening measures levels of a T-cell marker. This process uses the same dried blood spots already collected for screening of other disorders. Newborns with low levels of this marker are at higher risk for SCID. They need more testing to diagnose the condition.

Public health impact

Detailed data on how newborn SCID screening would affect public health were not available at the time of the report. However, newborn screening for SCID saves lives by allowing diagnosis and treatment early in life. Without screening, diagnosis can take 7 months or more after symptoms develop; some babies may never receive a diagnosis. Newborn screening allows diagnosis and treatment before symptoms arise.

Committee decision

The Committee voted in 2010 to recommend adding SCID to the RUSP. As of 2010, the RUSP recommends that state newborn screening programs include SCID.