Severe Combined Immunodeficiency (SCID):

What Healthcare Providers Need to Know

What is SCID?

Severe Combined Immunodeficiency (SCID) is a group of genetic disorders characterized by the absence or dysfunction of T lymphocytes and B lymphocytes. Newborns with SCID are at risk for severe, life-threatening bacterial, viral, and fungal infections. Without timely diagnosis and adequate treatment, patients with SCID do not survive infancy. SCID is a rare disease, with an estimated prevalence of 1 in 58,000 births.

What causes SCID?

SCID may be inherited in an X-linked recessive or autosomal recessive manner. The most common form of SCID is associated with a mutation in the IL2RG gene on the X chromosome and occurs almost exclusively in males. Another common type of SCID is caused by mutations in the gene that encodes adenosine deaminase (ADA). Other forms of SCID are caused by mutations in genes that code for the alpha chain of the IL-7R receptor, the Janus kinase 3 enzyme, RAG1 and RAG2, as well as other, less common mutations. Certain types of SCID are more common among specific groups, such as the Amish communities and Navajo Native American populations. Regardless of the genetic cause, all patients with SCID have a lack of T and functional B cells.

How is SCID identified?

SCID is identified using an assay to detect T-cell receptor excision circles (TRECs) which can be measured in dried blood spots obtained from newborns by heel prick. Newborns with SCID will usually have low or undetectable number of TRECs. The TRECs assay is also used to identify other, non-SCID conditions which are also characterized by T-cell deficiencies. As of December 2018, all states in the US have implemented newborn screening for SCID.

How to handle an abnormal newborn screen for SCID?

- Let the family know about the NBS result. Emphasize that a diagnostic test is needed to confirm whether the infant has SCID. Explain that the baby is at an increased risk of being diagnosed with an immune deficiency. As a precaution, the family should keep the baby in isolation.
- Consult with a pediatric immunologist who can provide more information on confirmatory testing.
- Do not give the baby vaccines before approval from an immunologist.
- Many mothers can carry cytomegalovirus (CMV) in their breastmilk. Tell mothers to stop breastfeeding until they consult with an immunologist and get tested for CMV.
- Recommend parents to use boiled water instead of bottled water with formula to ensure the baby does not get exposed to trace amounts of bacteria. Feeding bottles, including nipples, should also be boiled. Ready-to-feed formula, which does not require preparation or mixing, can also be used. Leftover formula should be discarded.
- If a SCID diagnosis is confirmed, refer the family to a pediatric immunologist with experience in treating SCID and to genetic counseling.
- Provide information about social or mental health services to the family.

How is SCID treated?

- Most infants with SCID are treated with hematopoietic stem cell transplantation (HSCT). The ideal donor is a sibling with identical human leukocyte antigens (HLA). If an HLA-identical sibling is not available, an unrelated donor or a parent could be used. Gene therapy has also been successful in treating some types of SCID, including ADA-deficient SCID, X-linked SCID, and Artemis SCID. Most gene therapies for SCID are currently in clinical trials. Treatments with enzyme and immunoglobulin replacement therapies are available for some forms of SCID, but they do not fully restore immune function.
- Until SCID patients undergo HSCT or gene therapy, isolation should be practiced, and supportive care with antibiotics and antifungals may be given to prevent infections. Because these preventive measures can only be used temporarily, children with SCID must undergo HSCT or gene therapy to restore the normal immune function. SCID patients who are diagnosed early and are infection free before HSCT or gene therapy, have a survival rate of about 95%.

Where to go for more information?

Research

- HRSA Newborn Screening Information Center
- Immune Deficiency Foundation: SCID Overview
- Immune Deficiency Foundation: TRECs and Newborn Screening
- SCID Compass: Low T Cell Counts and TRECs Screening
- NIH: Genetic and Rare Disease Information Center

Clinical Care

- PIDTC: Participating Clinical Centers
- Rotavirus Vaccine Guidance
- ACMG SCID ACT Sheet

Parent Support

- SCID Angels for Life Foundation
- SCID Compass: Informational Materials, Family Networking, and Advocacy Resources for Parents of Children Born with SCID
- SCID Compass: Parent Support Groups

Contact

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This project is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling $2.97 million with 0% financed with nongovernmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS or the U.S. Government.