Severe Combined Immune Deficiency Information for Health Professionals

Severe Combined Immune Deficiency (SCID) is a group of rare but potentially life-threatening, genetic disorders in which T lymphocytes fail to develop in addition to potential abnormal levels of B lymphocytes and NK cells. This “combined” impairment leads to life-threatening bacterial, viral, and fungal infections. The T cell receptor excision circles (TRECs) test is a screening test for a byproduct of normal T cell receptors. The TRECs test can identify SCID along with other disorders with low T cells such as DiGeorge syndrome and ataxia telangiectasia at birth.

Clinical Symptoms

The symptoms and severity of SCID vary between individuals based on the genetic defect. At birth, most newborns appear healthy, and they may be partially protected from the mother’s immunity. They may start to have common infections early on like otitis and bronchitis. In addition, infants may have thrush, chronic diarrhea, and failure to thrive. Within 2-4 months, patients often have opportunistic and potentially life-threatening infections such as pneumonia, sepsis, meningitis, and disseminated viral/fungal infections.

Incidence

In the United States, SCID occurs in approximately 1:40,000 to 1:50,000 live births but may be more common in certain populations where consanguinity is more common.

Inheritance Patterns

The most common form of SCID is the X-linked SCID and only affects males. However, there are autosomal recessive forms that affect both males and females. Parents of a child diagnosed with SCID may be unaffected. Parents who either have a child with SCID or another T cell lymphopenia immune deficiency disorder or who are known carriers of SCID should be referred to a medical geneticist or a genetic counselor for further discussion and testing.

Treatment

Newborn screening for SCID allows for earlier diagnosis and treatment. Studies have shown newborns who receive treatment, usually hematopoietic bone marrow transplantation, early have significantly improved outcomes. In fact, infants transplanted prior to 3 months of life have a 95% 20-year survival rate. Infants who are diagnosed later due to absence of newborn screening states often succumb to life-threatening infections, making transplantation much more complicated.

Screening Methodology

Testing methodology used in newborn screening to detect SCID and other T cell lymphopenias is done by testing for T cell receptor excision circles, otherwise known as TRECs. TRECs are discarded material when a T cell becomes activated leaving the thymus. If the TRECs are low or absent, then further testing is warranted to determine if the infant has a primary immune deficiency such as SCID or one of the other T cell lymphopenic disorders such as DiGeorge syndrome. TRECs can be low in premature infants and they may need repeat screening.

What to do After Receiving Presumptive Positive SCID Screening Results

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

1) Contact the family to inform them of the newborn screening result as additional tests are needed to determine if there is actually an immune deficiency.
2) If there is any evidence of infection, refer to Children’s Mercy or the University of Kansas Medical Center emergently for further evaluation and treatment.
3) If the infant requires blood products, ensure they are leukoreduced, irradiated, and CMV negative.
4) DO NOT give any live attenuated viral vaccines, which could cause serious illness. Avoid live viral vaccines in family members who have contact with the child.
5) Infants may not be able to be breastfeed depending on the CMV status of the mother.
6) Consult pediatric immunology, infectious disease, and hematology specialists immediately.
7) Provide the family with information about SCID and T cell lymphopenia.
8) Report confirmatory findings to the newborn screening program at 785-291-3363

REV. 1/27/2016
Confirmation of Diagnosis

Confirmatory testing may include repeating the newborn screening, particularly in premature infants, CBC with differential, absolute lymphocyte counts to determine presence or absence of particular lymphocyte (T, B, NK) subsets, lymphocyte functional assays, and genetic testing. The immunologist may order these tests and coordinate further care, and offer genetic counseling.

Communication of Results to Parents

If a baby has a presumptive positive SCID newborn screening result, additional testing needs to be performed to confirm a diagnosis. In accordance with Kansas Administrative Regulation 28-4-502, it is the responsibility of the attending physician or other birth attendant to obtain repeat specimens when needed to complete the screening process.

If a baby is diagnosed with abnormal TREC, the following points should be conveyed to parents:

- The baby needs to be followed at a center with pediatric immunology, infectious disease, and hematology.
- Further evaluation and treatment is urgent.
- Compliance with treatment is necessary for the best outcome.

For consultation contact:

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