Newborn Screening ACT Sheet
Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphopenia

Condition Description: Severe Combined Immunodeficiency (SCID) includes a group of rare but serious, and potentially fatal, inherited immune disorders in which T lymphocytes fail to develop and B lymphocytes are either absent or compromised. Impairment of both B and T cells leads to the term "combined." Untreated patients develop life-threatening, infections due to bacteria, viruses and fungi. The screening test for T cell receptor excision circles (TRECs), a byproduct of normal T cell development, identifies SCID as well as certain related conditions with low T cells. For example DiGeorge Syndrome with impaired thymus development may cause low T cells and low TRECs.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact the family to inform them of the newborn screening result. Point out that additional tests are required to determine whether the baby actually has an immune deficiency.
- Avoid exposing patient to illness pending completion of testing.
- If the infant has any signs of illness, refer to a pediatric hospital right away for evaluation, administration of immunoglobulin and antibiotics.
- If the infant requires transfusion of any blood product, be sure that only leukoreduced, irradiated products that are negative for cytomegaolovirus (CMV) are used.
- DO NOT give live attenuated rotavirus vaccine, which could cause serious diarrhea in a baby with SCID. This vaccine is to be given only after an immunology specialist confirms that the baby's immune system is normal.
- Consult with a specialist in pediatric immunodeficiency diseases (consult with a pediatric allergist/immunologist and/or infectious diseases specialist) who will assist with further testing.
- Provide the family with basic information about SCID and T cell lymphopenia (see resource list) and offer or arrange genetic counseling.
- Report confirmatory findings to newborn screening program.

Diagnostic Evaluation: Confirmatory studies include absolute lymphocyte counts, determination of the presence/absence of T and B lymphocytes and assessment of their function and molecular genetic testing.
The specialist will:

- Order diagnostic tests, likely to include: CBC with differential and lymphocyte subset enumeration.
- Coordinate further testing, antibody levels, lymphocyte proliferation to mitogens, and molecular genetic testing as deemed appropriate.
- Offer disease/genetic counseling

Clinical Considerations: Immunoglobulin infusions and prophylactic antibiotics are essential to protect against infections. Diarrhea, failure to thrive, otitis media, serious infections (pneumonia, meningitis and/or sepsis), and opportunistic infections commonly occur starting by 2-4 months of life in individuals with SCID. Oral thrush may be seen. Bone marrow hematopoietic cell transplantation may be curative, and outcomes are best if this is performed within the first 3 months of life or before infections occur. Enzyme replacement and experimental gene therapy are available for some SCID genotypes. The most common form of SCID is XSCID (X-linked SCID), occurring only in males. However, autosomal recessive forms of SCID affect both males and females. Specific gene diagnosis is important for directing therapy as well as providing genetic counseling.

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