Newborn Screening ACT Sheet

Severe Combined Immunodeficiency (SCID)

Differential Diagnosis: Other conditions involving immune system dysfunction such as DiGeorge Syndrome.

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 family to inform them of the positive SCID screening results, ascertain clinical status, and refer to a pediatric immunologist.
- If the infant has any signs of illness, contact a pediatric immunologist immediately.
- Well infants may stay at home safely during preliminary diagnostic testing as they have some protection from maternal antibodies. Families should be cautioned to avoid crowds and avoid contact with sick individuals.
- Infants with congenital or neonatal infections should be immediately evaluated by a pediatric immunologist.
- Avoid exposing patient to illness pending completion of testing.
- If the infant requires transfusion of any blood products, be sure that only leukoreduced, irradiated products that are negative for cytomegalovirus (CMV) are used.
- **The Primary Care Physician should obtain:** CBC with differential while awaiting the pediatric immunological consult. Pediatric immunology guidance prior to obtaining Chest X-ray.
- **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, a pediatric immunologist, who will assist with further testing.
- Provide the family with basic information about SCID and T cell lymphopenia

Repeat newborn screen if second screen has not been done.

Diagnostic Evaluation: Confirmatory studies may 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, immunoglobulin levels, lymphocyte proliferation to mitogens, and molecular genetic testing as deemed appropriate.
- These are very specialized tests and should only be performed in a laboratory with extensive pediatric immunology experience.
- Offer referral for genetic counseling to family when diagnosis confirmed.

Clinical Considerations: Immunoglobin 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 but may occur during the first weeks of life. Oral thrush may be seen. Hematopoietic stem cell transplantation (including bone marrow 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 etiologies. 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.

Additional Information:
March of Dimes @
http://www.marchofdimes.com/texas/getinvolved_4349.html
OMIM http://www.ncbi.nlm.nih.gov/omim
National Primary Immunodeficiency Resource Center
Jeffrey Modell Foundation http://jmfworld.org/
The American Academy of Allergy, Asthma & Immunology
http:///www.aaaai.org

SCID.net
The Immune Deficiency Foundation
http://primaryimmune.org/
American Academy of Pediatrics
http:///www.americanacademyofpediatrics.org
American College of Medical Genetics & Genomics
Genetic Home Reference
http://ghr.nlm.nih.gov/condition

Newborn Screening FACT Sheet
Severe Combined Immunodeficiency (SCID)

What is SCID?
SCID results when a baby's immune system does not work. Babies with SCID are not able to fight infection. They appear healthy at birth but can become sick very quickly when exposed to common illnesses. SCID is so rare that medical providers might not diagnose it until it is too late to provide lifesaving treatment.

Screening Tests
All Texas newborns get two blood tests that Screen's for 29 congenital disorders, and hearing screening. A positive newborn screen does not mean the baby has SCID, but it does mean the baby needs more testing to know for sure. The Primary Care Provider will be notified by the newborn screening program to arrange for additional testing.

Newborn screening will not detect all cases of SCID, and not all cases that screen positive for SCID will be diagnosed with SCID. If your child shows symptoms of SCID he/she should be evaluated by the specialist.

What Causes SCID?
SCID is a disorder that is passed on (inherited) from parents to child. One or both parents of an affected child carry a gene change that can cause SCID. Parents usually do not have signs or symptoms, or even know they carry the gene change. The genes that cause SCID prevent the immune system from maturing.

What Symptoms or Problems Occur with SCID?
Problems vary from child to child. These are some of the common ones:

- Frequent fevers
- Chronic skin infections
- Persistent rash
- Viral infections
- Chronic diarrhea
- Failure to thrive
- Thrush

Until the test results are back the baby should:

- Be kept at home
- Not be taken to daycare
- Use boiled tap water
- Avoid transfusions if possible. Discuss with pediatric immunologist before transfusions if unavoidable
- Babies with SCID should not receive live vaccines such as rotavirus, MMR, and Varicella Vaccine
- Avoid contact with other family members who have received a live vaccine for example FluMist®

What is the Treatment for SCID?
SCID can be treated. The usual treatment is bone marrow transplant.
Some babies who have a bone marrow transplant might still need lifelong treatment.

Medicines
Antibiotics to prevent infections
Gammaglobulin replacement (IVIG or SCIG)

Diet
Your baby should eat an age appropriate diet that is high in calories and protein. Discuss breast feeding with your pediatric immunologist. Do not use well water to mix baby's formula.

Things to Remember
Prompt treatment can improve a child's length and quality of life.
It is very important for the Primary Care Provider to give clear instructions to the parent regarding follow up testing and treatment.