

# Nebraska Newborn Screening Program

## Physician ACT sheet for POSITIVE SEVERE COMBINED IMMUNODEFICIENCY (SCID) Result

### Immediate Action Required:

- Contact Immunologist on call \_\_\_\_\_ Dr. Hana Niebur at 308-379-2949
- Contact the family to inform them of screening results and necessary follow up testing.
- Counsel parent(s) about avoidance of exposure to infectious disease while results of confirmatory testing are pending.
- If the baby has any signs of illness contact SCID team Immunologist.
- Do not give live attenuated virus vaccines including the rotavirus vaccine** until the baby is found to have a normal immune system. Household contacts should also not receive these vaccines.
- If the baby requires transfusion of a blood product **use only CMV negative, leukoreduced, irradiated products.**
- Interrupt Breast Feeding.** (Mom can pump and store breast milk.)

### Screening Test Results

Screening for SCID is done by counting T cell receptor excision circles (TRECs). TRECs are pieces of DNA that are by-products generated during normal T cell maturation in the thymus. Low numbers or absence of TRECs indicates the possibility of SCID or other conditions with low T cells. The screening test for SCID involves only TREC analysis and does not include testing for gene mutations for SCID.

### Condition Information

Severe Combined Immunodeficiency (SCID) is a group of rare, congenital, genetic conditions of the immune system characterized by failure of T cell development and absent or compromised B cells. Patients develop multiple and often

life-threatening infections which can be bacterial, viral or fungal. Infections commonly start at 2-4 months of age. Without treatment, patients with classic SCID will die early.

The most common form of SCID is X-linked and only affects males. There are also autosomal recessive causes of SCID that affect both males and females.

### Diagnostic Evaluation

**Contact the SCID team for guidance in arranging for immediate confirmatory testing.** A CBC and flow cytometry needs to be done. The flow cytometry will measure the numbers of types of T cells and their functional status. Further functional and molecular testing may be done depending on these results.

### Treatment

Newborn screening for SCID attempts to identify babies with SCID prior to any infections. **Bone Marrow Hematopoietic Cell Transplants have been found to be most effective if done prior to three months of life and before any infections occur.** Rapid diagnosis of SCID, allows the search for a donor to begin quickly.

Immunoglobulin infusions and prophylactic antibiotics are used to prevent infections. Some SCID genotypes may be treated with enzyme replacement or experimental gene therapy. Specific gene diagnosis is important for directing therapy and necessary for genetic counseling.

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