

Suggested Follow-up for Severe Combined Immunodeficiency (SCID)
Low TRECs

Possible Causes: Low levels of T cell receptor excision circles (TRECs) are associated with Severe Combined Immunodeficiency (SCID). Other conditions associated with low TRECs include reticular dysgenesis, coronin-1A deficiency and thymic aplasia/complete DiGeorge syndrome. T lymphocytes fail to develop and the affected infant may also have impaired B lymphocyte function.

Next Steps if Abnormal: **Potential medical emergency when TRECs are low and RNase P is within normal limits. The screening report will indicate Cq (Quantification Cycle) value instead of actual number of TRECs.** Cq is the number of cycles needed for the fluorescence of the amplified DNA to exceed the laboratory's established fluorescence threshold. **The Cq value of TRECs is inversely related to the copy number of TRECs in a specimen.** Specimens that have a low TREC content (low copy number) have a higher Cq value, because the specimens have to go through more thermal cycles to achieve fluorescence above the baseline.

See infant as soon as possible to ascertain health status. Consult pediatric specialist (immunology or pediatric infectious disease) and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include specialized flow cytometry and molecular testing to determine specific mutations. In addition, repeat TREC on filter paper and send to the DHEC laboratory. Low TRECs with low RNase P may indicate DNA amplification failure. Prompt repeat screening is necessary to rule out SCID in these infants.

Neonatal Presentation: Usually none. Median age for onset of symptoms is 8 weeks of age.

Emergency Treatment: Usually none.

Standard Treatment: Bone marrow transplantation by 3 months of age is associated with the best outcomes for SCID. Infants with other conditions may be treated with medications.

Advice for Family: Provide basic information about SCID and related immunodeficiencies. The handout, *When Baby Needs a Second Test for an Immune System Disorder*, may be used for this purpose. **Parents should be instructed to avoid exposure of the infant to anyone with viral/bacterial illnesses until SCID is definitively ruled out.** No vaccines should be given until cleared to do so by the specialist. The specialist may advise breastfeeding mothers to suspend breastfeeding while their blood is checked for anti-CMV IgG antibodies and CMV DNA. These mothers should be encouraged to pump and freeze their breast milk during this time. Prompt resumption of breastfeeding is encouraged if the mother is seronegative. **Only leukoreduced, CMV negative, irradiated blood should be used if a transfusion is necessary.**

NOTE: Premature infants may have low TRECs due to immaturity of the immune system. Prompt repeat screening is indicated. The pediatric specialist (immunology or pediatric infectious disease) may recommend flow cytometry if TRECs are low in a second blood spot specimen. Low TRECs may also be found in specimens obtained from infants who have undergone thymectomy/cardiac surgery if the specimen is collected after surgery.

Internet Resources: <https://www.acmg.net/StaticContent/ACT/SCID.pdf>
<http://primaryimmune.org/>
<http://jmfworld.org/>
<http://www.niaid.nih.gov/topics/immunodeficiency/Pages/Default.aspx>