

SCID FACT SHEET FOR PRIMARY CARE PROVIDERS

NEWBORN SEVERE COMBINED IMMUNODEFICIENCY (SCID) SCREENING

Early diagnosis and treatment can correct the immune system malfunction of infants with SCID and vastly improve their survival. This is a fatal disorder unless recognized and treated before life-threatening infections develop.

- Implementation in North Carolina in 2017
- **SCID Screening Test**
 - Uses the same dried blood samples already collected from newborns;
 - The TREC test is an assay that detects the number of T-cell Receptor Gene Excision Circles, or TRECs, that are produced during normal T-cell maturation, but that are *absent or severely reduced* in infants with SCID;
 - The TREC test is very sensitive and has a false positive rate well below 1%;
 - The TREC test does not diagnose SCID and detailed guidance will be given to the infant's primary care provider when there are abnormal results.
- **Conditions found by screening for low or absent TREC:**
 - Typical SCID, due to defects that include IL2RG (X-linked), ADA, IL7R, JAK3, RAG1, RAG2, DCLRE1C (Artemis), TCRD, TCRE, TCRZ, and CD45;
 - Leaky SCID or Omenn syndrome, due to mutations in typical SCID genes that do not completely abolish gene function;
 - Variant SCID, with persistently low T-cells but no defect in a known SCID gene;
 - Other conditions such as Di George Syndrome, Ataxia-telangiectasia and Trisomy 21.
- **Follow-up of an abnormal screen**
 - Detailed recommendations will be provided to the primary care provider with the newborn screening result that **may** include but are not limited to:
 - Immediate evaluation of the infant by the primary care provider;
 - Instructions to share with parents regarding precautions necessary to avoid risk of infection until the evaluation is completed (i.e. boiling tap water, avoiding live vaccines);
 - Prompt referral to a Pediatric Immunologist for diagnostic testing and further evaluation.

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- **Immunologists**
 - **Carolina's Medical Center, Charlotte, NC**
Phone: (704) 381-6803
Immunologist on call: (704) 355-4088 ext. 8125
 - **Duke University Medical Center, Durham, NC**
Phone: (919) 684-3204
Immunologist on call: (919) 970-7311
 - **University of North Carolina School of Medicine, Chapel Hill, NC**
Phone: (919) 962-5136, option #4 (administrative specialists Jessi Stark and Samantha Kemper)
 - **Wake Forest University School of Medicine, Winston Salem, NC**
Phone: (336) 716-4843
After normal business hours, contact the Wake Forest Immunology Fellow/Attending on Call via the Physician Access Line (PAL) at (336) 716-7654

- **Resources for more information**
 - Newborn Screening (NBS) Follow-up Coordinator
(919) 707-5634
<http://slph.state.nc.us>
 - Immune Deficiency Foundation for SCID
<http://primaryimmune.org/treatment-information/newborn-screening/>
 - National Human Genome Research Institute of the NIH
<https://www.genome.gov/13014325>
 - Centers for Disease Control and Prevention
<http://www.cdc.gov/newbornscreening/scid.html>