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](http://slph.state.nc.us)
  - Immune Deficiency Foundation for SCID  
    [http://primaryimmune.org/treatment-information/newborn-screening/](http://primaryimmune.org/treatment-information/newborn-screening/)
  - National Human Genome Research Institute of the NIH  
    [https://www.genome.gov/13014325](https://www.genome.gov/13014325)
  - Centers for Disease Control and Prevention  
    [http://www.cdc.gov/newbornscreening/scid.html](http://www.cdc.gov/newbornscreening/scid.html)