



Materials for Parents Following Newborn Screening



*Made possible by a grant from the
Association of Public Health Labs*



Mission Statement

Founded in 1980, the Immune Deficiency Foundation is the national patient organization in the United States dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research



IDF SCID Initiative

Mission

- To expand public awareness and treatment options for SCID
- To increase the probability of early detection and treatment
 - Goal: Advocate for screening in all 50 states
- To fund research efforts for treatments, therapies, and ultimately a cure for SCID
- To provide educational events for individuals and families affected by SCID



History of SCID Screening

- In 2003, The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was established and since then, IDF has advocated for SCID screening
- In 2007, IDF presented, testified and submitted letters advocating that SCID be added to the recommended newborn screening care panel
- In 2009, IDF completed the first national survey of parents of children with SCID, published in 2011, which found SCID babies tested at birth are much more likely to survive than those identified later
- In 2010, the Secretary of the Department of Health and Human Services, added TREC screening for SCID to the core panel for all newborns in the United States



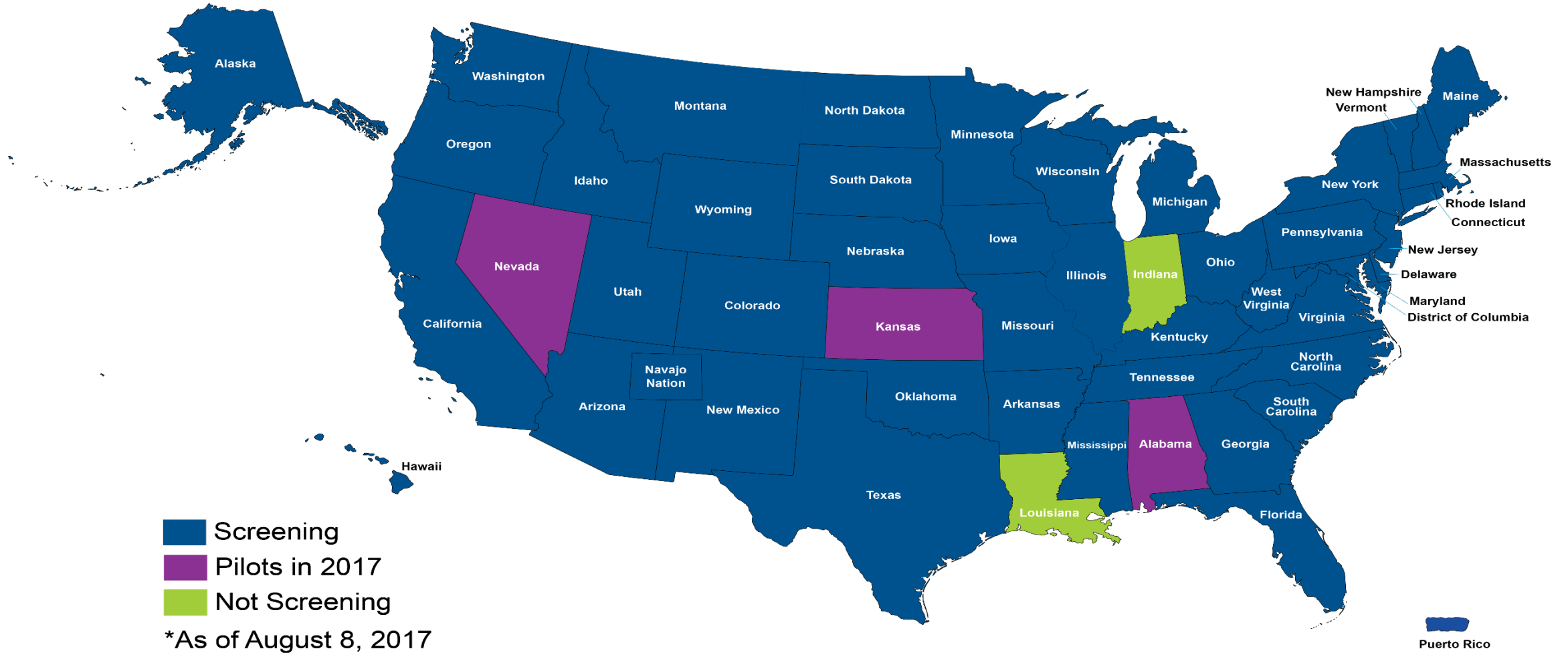
Newborn Screening for SCID

- 44 states as well as Puerto Rico, Washington, D.C. and the Navajo Nation of Arizona are screening
- 90% of all births in the United States are being screened for SCID
- 4 states have pilots or are committed to begin screening
- 2 states still left to commit to fully screen



SCID Newborn Screening: Current Status of Implementation Map*

45 States Currently Screening for SCID - 90% of all newborns in the U.S. are receiving SCID screening



2008	2009	2010	2011	2012	2013	2014	2015	2016	2017
Wisconsin	Massachusetts	California	Delaware	Colorado	Minnesota	Illinois	Arkansas	Alaska	Missouri
		New York	Michigan	Connecticut	Ohio	Iowa	Hawaii	Georgia	North Carolina
				Florida	Utah	Maine	New Hampshire	Idaho	Arizona
				Mississippi	Wyoming	Nebraska	Oklahoma	Kentucky	
				Texas		New Jersey	South Carolina	Maryland	
						Oregon	South Dakota	Montana	
						Rhode Island	Virginia	New Mexico	
						Washington		North Dakota	
						Washington, DC		Pennsylvania	
						West Virginia		Puerto Rico	
								Tennessee	
								Vermont	

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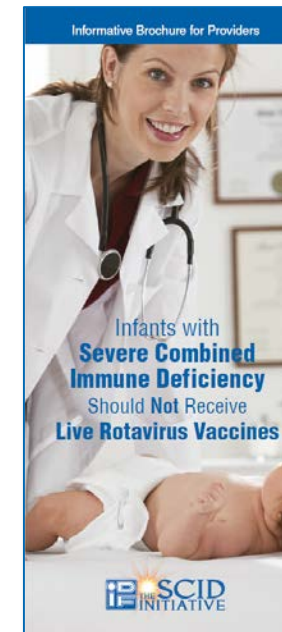
Screening and Early Treatment Saves Lives

- If detected and treated within the first four months of life, there is a 94% chance of a child going on to live a healthy, productive life
- Without early detection and appropriate treatment, a baby with SCID will likely not see his or her first birthday
- If an infant survives, the child will likely become severely disabled due to complications from SCID, requiring additional medical costs for the rest of their life



SCID Resource Development 2010-2017

- [SCID Newborn Screening Campaign webpage](#)
- Survey of all state health departments regarding the process for adding condition to the newborn screening panel
- SCID Newborn Screening blog to share volunteer activities
- Development of SCID Newborn Screening Toolkit
- Development and Distribution of:
 - A Guide for Parents Following a Diagnosis flyer
 - Abnormal Screening flyer
 - Rotavirus Vaccine brochure
 - Understanding the Low T Cell Results from Your Baby's Newborn Screening brochure – funded through APHL



SCID Activities 2016-2017

- Supported full implementation of SCID screening in 12 states
- Presented to professionals and leaders at AZ SCID awareness week
- Worked directly with AZ Governor's office to advocate for implementation
- Supported volunteers and other advocacy organizations in Arizona, Louisiana and Indiana to remove legal and fiscal barriers
- Developed slide presentation to educate families impacted by SCID
- Developed low T cell brochure for labs to provide to families whose children do not have SCID but do have low lymphocytes



TREC Test

- The T-cell Receptor Exclusion Circles (TREC) test can detect SCID from the same blood spot collected to screen for a variety of conditions
- Recommended by the U.S. Department of Health and Human Services (HHS) and the Centers for Disease Control and Prevention (CDC) as the preferred way to screen newborns for SCID
- Identifies other congenital conditions including DiGeorge syndrome, trisomy 21, ataxia telangiectasia and CHARGE syndrome
- Identifies infants with low lymphocytes (low T cells) which may be a result of conditions including vascular leakage, neonatal leukemia, gastrointestinal malformations and intrauterine growth retardation



Understanding the Low T Cell Results: A Guide for Parents

- What does a low T cell result mean from your baby's newborn screening?
- What is TREC testing?
- Why is evaluation and continued follow up by a specialist important?
- What conditions or disorders can be seen in babies with no or low T cells?
- Are low T cells common in babies?
- How many babies with abnormal TREC test results have low T cells not associated with SCID?
- How are the tests completed?
- Could your baby be diagnosed with SCID at a later date?
- What should you do to keep your baby healthy while awaiting a diagnosis?



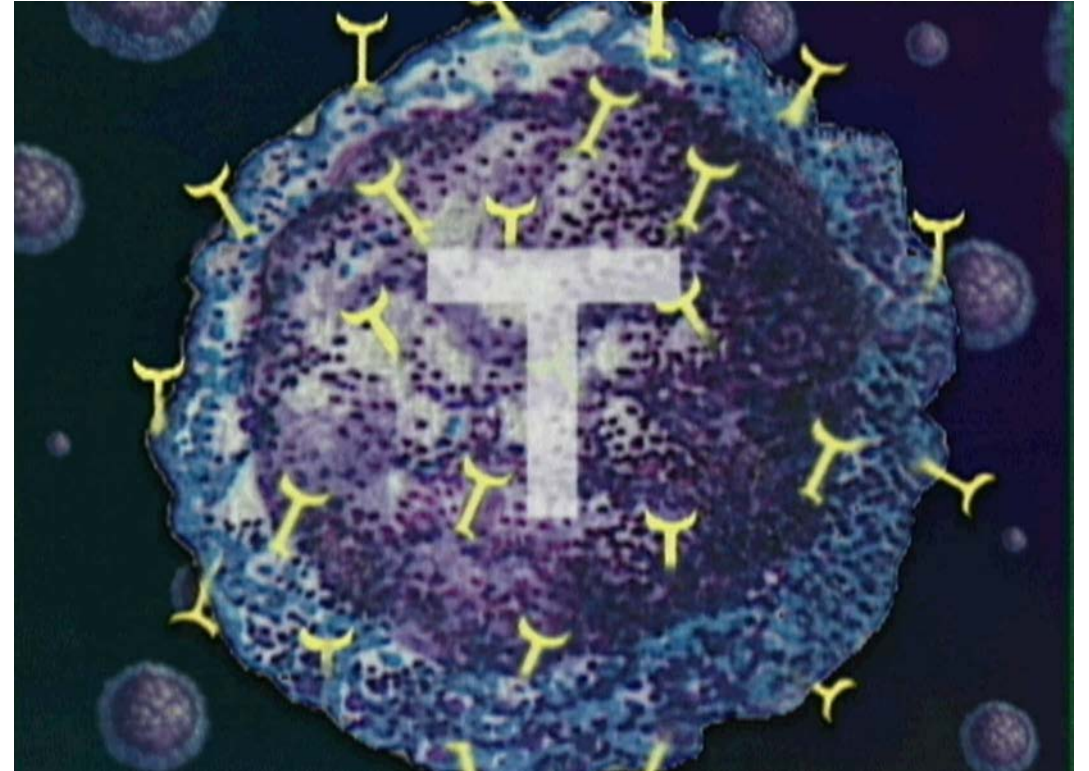
What Conditions or Disorders can be Seen in Babies with No or Low T Cells?

- Severe Combined Immune Deficiency (SCID)
- Combined immunodeficiency (leaky SCID)
- Congenital abnormalities associated with low T cells
- Prematurity
- DiGeorge Syndrome
- Genetic syndromes associated with low T cells
- Lymphopenia (low lymphocyte counts) of infancy
- Other primary immunodeficiencies
- Secondary T cell deficiency loss of T cells
- Transient immunosuppression from maternal medication or certain viral exposures during pregnancy



Low T Cell Statistics

- Approximately 1 in 20,000 babies have low T cells based on the data from newborn screening.
- 40-50% of babies with an abnormal TREC test have low T cells. Of these, about two thirds will have low T cells not associated with SCID.



How to Keep Your Baby from Getting Sick While Awaiting Diagnosis

- Ask your immunologist what to do while your child is still be evaluated
- Ask your immunologist whether to breastfeed or formula feed
- Maintain good hygiene, including frequent hand sanitizing
- Avoid contact with sick people
- Maintain communication with your immunologist and pediatrician



Severe Combined Immune Deficiency (SCID): A Guide for Parents Following a Diagnosis

- What is SCID?
- What do I do now?
- Safety precautions
 - Isolation
 - Avoid live virus vaccines
 - Blood or platelet transfusions must be irradiated
- How is SCID treated?
- What causes SCID?



Isolation While Awaiting Transplant

- While infants can be successfully treated with bone marrow or stem cell transplants, the procedure may need to be delayed for many reasons
- A newborn with an immune disorder may not appear sick because the mother's antibodies provide protection in the first few months of life
- While awaiting transplant, parents must ensure that the child is isolated and take precautionary measures



Treatment while in Isolation

- Preventive antibiotics, generally given by mouth, may be needed to ward off infections
- Infants may be given replacement immunoglobulin through the vein or under the skin to provide more protection against infections



Precautions for Parents of Newborns with SCID

- Isolate baby from anyone with a cold or infection
- Avoid crowded public places
- Avoid children who are not members of the immediate family
- Follow strict hand washing before touching your baby
- Inform physician about family pets



Avoid Live Virus Vaccines: Informative Brochure for Providers

- Although recommended for healthy babies, vaccines such as rotavirus, chickenpox, mumps, measles and live poliovirus **should not** be given to any baby suspected of having a poorly functioning immune system.
- Other household members **should not** receive any live vaccines



Treatment

- The most common treatment for SCID is a bone marrow or stem cell transplant from a healthy donor.
- The ideal donor is a perfectly matched sibling
- Excellent results are possible with a related donor, such as a parent or unrelated donor found to be a good match
- In some instances, other treatments are possible
- All treatment options require a specialized medical center where there are physicians (pediatric immunologists) with experience treating SCID



SCID Isolation Video

- *SCID – A families journey through Isolation* was created by IDF featuring a real family with a baby with SCID
- Answers questions that families have, highlights a success story and provides tools to help others cope with the challenges of SCID
- Funded through APHL Grant
- Posted on the IDF Advocacy Channel in December 2015 and as of August 2, 2017 has 9734 views

- **Video:**

<https://www.youtube.com/watch?v=YBAPd5VQyfl&list=PLF94D72EBA3DC58E1&index=6>

<https://www.youtube.com/user/PrimaryImmune>



Questions

