



Newborn Screening for Severe Combined Immune Deficiency: Advocacy, Challenges, and Next Steps

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Mission Statement

Founded in 1980, the Immune Deficiency Foundation (IDF) 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.



www.primaryimmune.org

Reach & Impact of IDF Programs – 2010

- Nearly **225,000** visits to IDF websites
- **18,000** copies of the Advocate Newsletter mailed three times a year, thousands more handed out at medical and patient meetings
- Over **10,000** E-Newsletters sent monthly
- Approximately **10,850** patients and healthcare professionals attended more than **135** educational programs (in **111** cities, **41** states, **3** countries)
- Over **165,000** IDF education and awareness pieces were mailed
- IDF addressed over **8,500** direct patient contacts

2009 IDF Survey of Severe Combined Immunodeficiency: Experiences with Diagnosis & Treatment



IDF presented data before SACHDNC February 26, 2009 supporting the addition of SCID to the Newborn Screening Panel which has since been published in *CLINICAL IMMUNOLOGY* online Oct. 2010, and currently in print for Jan. 2011

“Early vs. Delayed Diagnosis of Severe Combined Immunodeficiency: A Family Perspective Survey”

**Alice Chan, MD, PhD; Christopher Scalchunes, MPA;
Marcia Boyle, MS; Jennifer Puck, MD**

Major Findings from the survey:

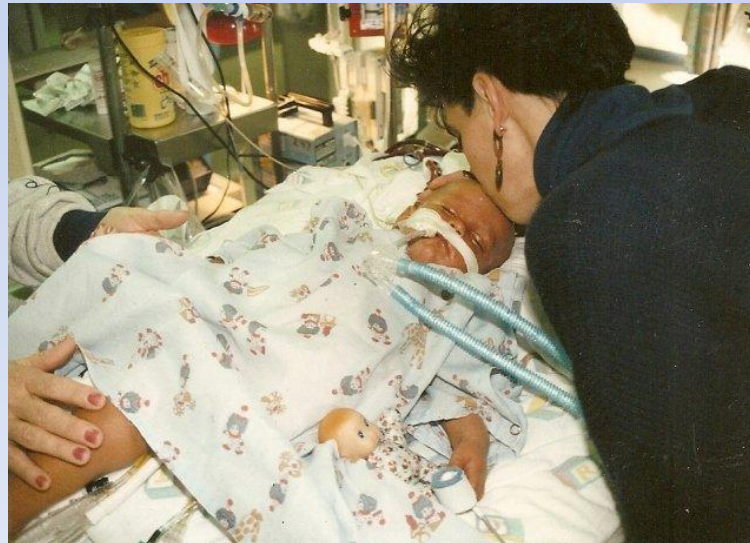
- Morbidity, delayed diagnosis, increased medical costs and death were potentially avoidable outcomes that might have been prevented had a universal newborn screening test been in place
- Our study design had the unique ability to include a more complete ascertainment of the SCID population than is included in studies from referral centers. The study suggests that approximately half of the deaths among infants with SCID may be missed in the statistics arising from referral centers.

IDF SCID Newborn Screening Campaign



Following the recommendation of HHS Secretary Sebelius that all states include SCID in newborn screening panels, IDF launched the IDF SCID Newborn Screening Campaign

Goal: The Campaign goal is to have SCID included in newborn screening protocols in all 50 states and territories through Advocacy and Education



Current Status of Implementation



- **States and Territories Currently Screening for SCID:**

Wisconsin

Massachusetts

California

New York

Louisiana

Puerto Rico

Texas* - limited pilot program in select hospitals, screening 20,000 of the states' newborns

- **States where Newborn Screening Advisory Committees have voted to recommend the addition of SCID, but screening has not yet begun:**

Colorado

Iowa

Minnesota

Michigan

Delaware

Rhode Island

North Carolina

IDF Advocacy Activities



- Surveyed all state health departments regarding process for adding condition to the newborn screening panel
- Met with state health departments – provided resources from cost analysis figures to expert immunologists in the state to contact for follow-up on positive screens
- Formed alliances with other advocates in the state to collaborate on NBS – March of Dimes in Georgia, Pennsylvania Medical Society
- Supported volunteer activities in the states

IDF Advocacy Activities



IDF staff and volunteers have engaged in advocacy activities in approximately **30 states**

- Five state Advisory Committees have voted to recommend SCID screening at meetings IDF volunteers or staff presented
 - Delaware, Michigan, Minnesota, Rhode Island, North Carolina
- IDF staff and volunteers have worked in the following states with activities ranging from presentations to Advisory Committees to providing data and resources to discussions with Department of Health staff
 - Connecticut, Florida, Ohio, Georgia, Oregon, Illinois, Pennsylvania, Maryland, Utah, New Jersey, Virginia, Washington

IDF Educational Activities



- Launched IDF SCID Newborn Screening Campaign webpage information for advocacy
- Developed IDF SCID Newborn Screening Advocacy Toolkit for use in educating policymakers
- Posts to the SCID Newborn Screening blog to keep our community aware of volunteer activity and opportunities
- Created and distributed of IDF SCID Live Rotavirus brochure to warn providers about the dangers of administering the vaccine to infants with SCID
- Presented to all 50 state lab programs – CDC meeting, Atlanta, GA on October 28, 2010
- Produced of two video stories from SCID parents emphasizing the importance of early detection – one shared at CDC meeting and both are on IDF NBS Campaign website

For more information:

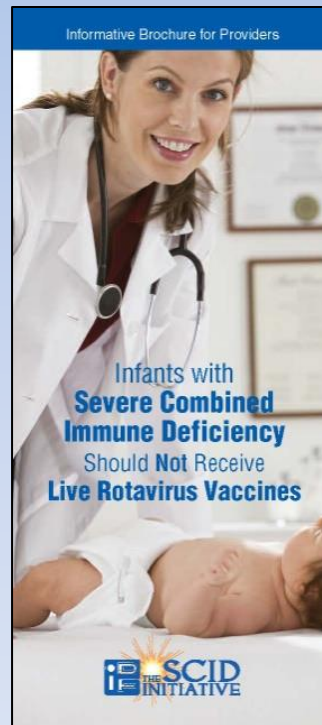
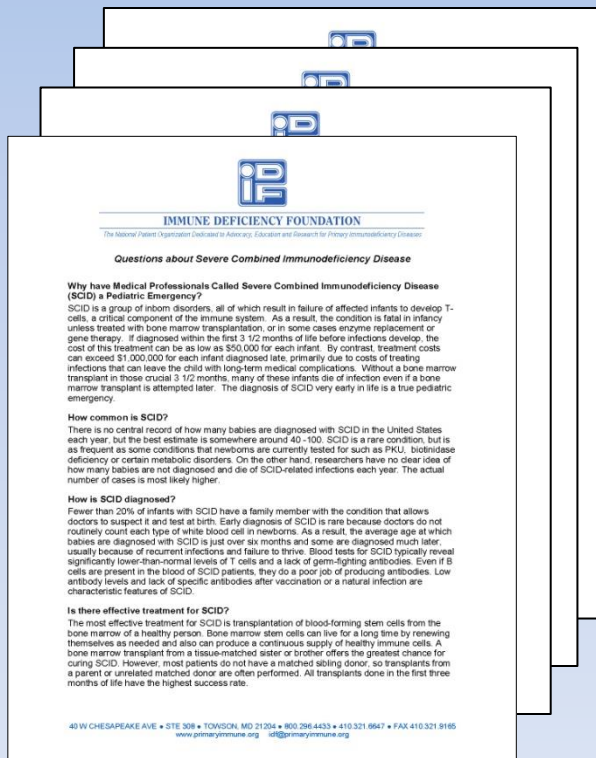
https://www.primaryimmune.org/advocacy_center/scid/scid_newborn_screening_initiative.asp

IDF Patient Educational Materials and Other Resources for SCID

IDF SCID Newborn Screening Toolkit

IDF SCID Live Rotavirus Brochure

IDF SCID Newborn Screening Blog



https://www.primaryimmune.org/advocacy_center/scid/scid_newborn_screening_initiative.asp

IDF SCID Newborn Screening Videos

Video stories from SCID parents emphasizing the importance of early detection – one shared at CDC meeting and both are on IDF NBS Campaign website



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Challenges to Implementation

- Funding – some states have identified funding as the main barrier against implementation as cost estimates for developing the infrastructure of SCID screening are between \$500,000 and \$1 million to the state
- Waiting our turn - Prior commitment to other disease groups that must be implemented before SCID screening can begin
- Follow-up – Need for experts in primary immunodeficiency for diagnosis and treatment following a positive screen
- Current lack of an FDA approved assay for the screening

IDF Recommendations

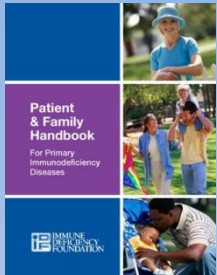
- States must develop networks of specialists in primary immunodeficiency for diagnosis and treatment of a given patient
- States must develop strategies that assure patients access to specialists in primary immunodeficiency diseases including sending patients out of state to medical centers with expertise in bone marrow transplantation for SCID when such resources are not available in the state.
 - Some state Medicaid programs are reluctant to approve a patient going out of state even if there are no SCID specialists in its state.

Educational Needs Within the State



States need to develop systems to educate and communicate next steps to physicians and families

- Distribution of educational piece for pediatricians following identification of a positive test result
 - Where to go for a diagnostic test and evaluation of the most appropriate therapy – referral to a specialist in primary immunodeficiency
- Educational piece for parents who receive a positive screen
 - What it means, what to do next to receive a diagnosis
- Educational piece for parents who receive a definitive diagnosis



Patient & Family Handbook Chapters on SCID and Treatment of SCID



Severe Combined Immunodeficiency

CHAPTER 5

Severe Combined Immunodeficiency is an uncommon primary immunodeficiency in which there is combined absence of T-lymphocyte and B-lymphocyte function. There are a number of different genetic defects that can cause Severe Combined Immunodeficiency. These defects lead to extreme susceptibility to very serious infections. This condition is generally considered to be the most serious of the primary immunodeficiencies. Fortunately, effective treatments, such as bone marrow transplantation, exist that can cure the disorder and the future holds the promise of gene therapy.

Topics on SCID include:

- Definition
- Clinical Presentation
- Diagnosis
- Inheritance
- General Treatment
- Expectations



CHAPTER 18

Specific Medical Therapy

There are several specific medical therapies available for patients with primary immunodeficiency diseases. Effective therapies for these disorders are a reality for most patients, improving their productivity and allowing most of them to lead active lives, pursue careers and have families.

- ## Specific Therapies Described Include:
- Immunoglobulin Therapy
 - Hematopoietic Stem Cell Transplantation
 - PEG-ADA
 - Gene Therapy

Recognizing the Needs of Patients

Developing educational materials for families who receive a positive screening result

- IDF is working with a specialist in primary immunodeficiency to develop a “brochure” that will educate parents who have received a positive screen and emphasize the importance of confirmatory testing
- Will explain what SCID and other T-cell lymphocyte deficiencies are and appropriate treatment
- Relieve concerns by explaining what to do next – doctor will contact for additional testing
- Give links to additional resources on SCID and other T-cell lymphocyte deficiencies

THANK YOU!