IMMUNE DEFICIENCY FOUNDATION

The National Patient Organization Dedicated to Advocacy, Education and Research for Primary Immunodeficiency Diseases

Immune Deficiency Foundation praises ACHDNC recommendation for SCID screening in newborns

Washington, D.C. – On January 21, 2010, the Advisory Committee on Heritable Disorders in Newborns and Children voted unanimously to add screening for Severe Combined Immune Deficiency or SCID - commonly known as bubble boy disease — to the core panel for universal screening of all newborns in the United States.

SCID is a primary immunodeficiency; affected infants lack T lymphocytes, the white blood cells that help resist infections due to a wide array of viruses, bacteria and fungi. Babies with SCID appear healthy at birth, but without early treatment, most often by bone marrow transplant from a healthy donor, these infants cannot survive.

The Advisory Committee's policy recommendation will now be presented to Kathleen Sebelius, Secretary of Health and Human Services. Ms. Sebelius has 180 days to consider and respond to the committee's proposal.

Jennifer Puck, Professor of Pediatrics at the University of California, San Francisco and an expert in the field, nominated SCID for consideration by the Committee after pioneering development of a test that can detect SCID in the dried blood spot filter cards that are currently collected from all babies to screen for a variety of inborn disorders.

"The Advisory Committee has taken a momentous step forward for the primary immunodeficiency community," said Marcia Boyle, President & Founder of the Immune Deficiency Foundation (IDF), the national patient organization for persons with primary immunodeficiency diseases. "The IDF has strongly supported and worked tirelessly toward this goal for many years. But it is imperative that we sustain this momentum by working to establish newborn screening programs in all 50 states."

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was chartered in February 2003 to perform evidence-based reviews and advise the Secretary regarding application of new screening tests, technologies, policies, guidelines and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk for, heritable disorders. SCID is the first new disease to be recommended for addition to the federal uniform core-screening panel by the evidence-based Committee review process.

"Although this recommendation has been in development for two years," said Dr. Amy Brower, parent, researcher and former ACHDNC committee member, "and it may take several more years to implement screening in all 50 states and US territories, we must work to reduce the time from this recommendation to the widespread adoption of testing and treatment."

In 2004, Duke University Professor of Pediatrics and Immunology, Dr. Rebecca H. Buckley – currently the chair of the Immune Deficiency Foundation's Medical Advisory Committee and an internationally known expert on SCID– testified before the ACHDNC (to which she was subsequently appointed in 2007).

Dr. Buckley informed the panel that SCID is a pediatric emergency. If a baby with SCID receives a bone marrow transplant in the first 3.5 months of life, the survival rate can be as high as 94 percent. However, the survival rate drops to less than 70 percent for infants who are transplanted after that age. The main causes for the drop in survival rate are serious infections babies with SCID develop prior to transplantation.

"As the parent of a child who was diagnosed with SCID only after he became critically ill," said Barb Ballard, a member of the IDF Board of Trustees, "I have to say I am immensely pleased with the recommendation of the committee."

In addition to the IDF, the Jeffrey Modell Foundation and members of the public whose children were affected with SCID argued in favor of SCID screening. One child born in Wisconsin, where pilot SCID screening is ongoing, was diagnosed at 9 days of age and successfully cured by timely bone marrow transplantation, while another child born in Oregon, where screening does not take place, developed failure to thrive and multiple infections and did not survive.

"Our goal is to have Newborn Screening for SCID passed in *all* 50 states," said Heather Smith, co-founder of SCID Angels for Life, who lost her six-month-old son, Brandon, to this devastating disease.

"We have already lost way too many children to this disease," Ms. Smith concluded, "and newborn screening is the only way we can put an end to the pain and suffering these children are forced to endure."

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The Immune Deficiency Foundation is the national patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research. For information on this media release, please contact Marcia Boyle, president, at 410-321-6647 or idf@primaryimmune.org. For further information please visit the IDF website at www.primaryimmune.org, or call IDF at 800-296-4433.