

Hyper-IgE syndrome, among others. She has been part of gene therapy development for patients with X-linked SCID who failed bone marrow transplantation and now Artemis-deficient SCID. Noting that early diagnosis with treatment prior to occurrence of infectious complications is the key to successful outcomes for SCID patients, she conceived and developed a test to screen newborns for SCID using PCR quantitation of T cell receptor excision circles (TRECs) in DNA isolated from infant dried blood spots. TREC testing, now part of the standard newborn screening panel in several states, allows infants affected with SCID and other T cell lymphopenic disorders to be detected early and treated. ●

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JENNIFER PUCK, M.D.

EARLY DETECTION OF SEVERE IMMUNE DEFECTS
BY NEWBORN SCREENING

DECEMBER 12, 2013
4:00 P.M.
208 LIGHT HALL

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Upcoming Discovery Lecture:

DAVID M. SABATINI, M.D., Ph.D.
Whitehead Institute for Biomedical Research

January 9, 2014
208 Light Hall / 4:00 P.M.

VANDERBILT  UNIVERSITY
MEDICAL CENTER

EARLY DETECTION OF SEVERE IMMUNE DEFECTS BY NEWBORN SCREENING

Background

Newborn screening (NBS) for severe combined immunodeficiency (SCID) using T cell receptor excision circles (TRECs) was initiated in Wisconsin in 2008, and added to the DHHS Secretary's Recommended Uniform Panel of newborn screened diseases in May 2010. Through May, 2013, 15 states and the Navajo Nation had adopted universal SCID NBS, with additional states planning to do so.

Objective

To assemble data reflecting the spectrum and experience of state SCID NBS programs.

Methods

State public health laboratory professionals and state-designated immunologists were invited to submit SCID screening algorithms, overall test performance data and de-identified clinical and laboratory information regarding total cases screened and cases with non-normal results. Numbers of infants with SCID and other diagnoses were classified according to standard criteria, and immunological interventions were recorded.

Results

Ten states and the Navajo Nation participated, representing a total of 3,031,883 infants screened with a TREC test. Diagnoses included 52 cases of SCID disorders, typical SCID, leaky SCID and Omenn syndrome, affecting 1 in 58,000 infants. There were 398 additional cases of T cell lymphopenia, or 1 in 7,700 infants. The infants identified received immune restoring interventions including hematopoietic cell transplantation, thymus transplantation, enzyme replacement therapy and gene therapy, as well as protective interventions including immunoglobulin infusions, antibiotics, transfusion precautions, and live vaccine avoidance.

Conclusions

NBS successfully identified SCID cases early in life, making possible prompt intervention previously shown to optimize survival. The experience of states already performing SCID NBS will facilitate adoption by additional states in the U.S. as well as by other countries.



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Dr. Puck earned a medical degree at Harvard Medical School Health Sciences and Technology Program and completed a residency in pediatrics at Washington University Medical Center in St. Louis, Mo., followed by fellowships in infectious diseases and immunology at Baylor College of Medicine in Houston. After faculty appointments at the University of Pennsylvania Medical School and the Division of Intramural Research, National Human Genome Research Institute, NIH, she joined UCSF in 2006 as Professor of Pediatrics. She was founding Chair of the National CTSI Consortium Child Health Oversight Committee and serves on the Medical Advisory Committee of the Immune Deficiency Foundation, the Committee on Primary Immunodeficiency Disease of the International Union of Immunological Societies and the Pediatric Scientist Development Program. She has been elected to the American Society of Clinical Investigation (ASCI), Society for Pediatric Research (SPR), Association of American Physicians (AAP), American Pediatric Society (APS) and Institute of Medicine. In 2012 she won the Abbot Award from the American Society of Microbiology in 2013.

In addition to her patient care as an immunologist and teaching biomedical trainees at all levels, Dr. Puck has a basic and translational research program that focuses on human primary immunodeficiencies as well as mouse models of lymphocyte development. Her scientific contributions include mapping and identification of the genes for X-linked severe combined immunodeficiency (SCID) and autoimmune lymphoproliferative syndrome (ALPS), and Job's

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