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WI Public Health Lab Identifies First SCID Baby Using New TREC Assay

Silver Spring, MD—The Wisconsin State Laboratory of Hygiene at the University of Wisconsin-Madison has identified the first baby with classical Severe Combine Immune Deficiency (SCID) as part of their newborn screening program. SCID babies, if undiagnosed, eventually develop severe life-threatening infections with a 100% mortality rate, usually within the first year of life. With prompt diagnosis and treatment before the infections, SCID is curable when treated by hematopoietic stem cell transplantation (HSCT). This is the first and only condition on the newborn screening panel of 30 genetic diseases that is curable if diagnosed very early in an infant's life. SCID refers to a collection of inherited immunodeficiencies characterized by profound defects of both T cell and B cell arms of the immune system.

According to Dr. Christine M. Seroogy, associate professor in the Department of Pediatrics at the University of Wisconsin, "The ability to cure validates why we do this test. It is working the way it was designed to work."

In an extraordinary collaborative effort in 2007, Dr. Mei Baker, science director of the Wisconsin State Laboratory of Hygiene's Newborn Screening Program, worked closely with Drs. John Routes and William Grossman, both with the Children's Hospital of Wisconsin and Medical College of Wisconsin, to develop a molecular assay that detects the absence of TRECs (T cell Receptor Excision Circles) using dried bloodspots and determined that it was the most effective way to diagnose SCID. TRECs are small pieces of DNA generated in T cells as they mature.

"This is the first time a molecular assay is being used as a primary screening test in newborn screening," says Dr. Baker, who is also an assistant professor of pediatrics at the University of Wisconsin. "The SCID test is a perfect example of applying advanced molecular knowledge and technology in public health newborn screening programs."

Dr. John M. Routes, medical director, section of allergy and immunology, Children's Hospital of Wisconsin feels that newborn screening for SCID has already been a tremendous success. "In 2007 we determined that it was feasible to conduct high throughput TREC assays for SCID screening within a public health laboratory and now it

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is happening and has already saved a baby's life.” Dr. Routes was responsible for securing initial funding for the project, a \$250,000 grant from the Jeffrey Modell Foundation and a \$250,000 grant from the Children’s Hospital of Wisconsin.

Since then, a grant from the Centers for Disease Control and Prevention (CDC) has allowed the program to continue. Part of the grant from CDC was to be used to train laboratories in other states to perform the TREC assay. The Wisconsin State Laboratory of Hygiene as well as the New England Newborn Screening Program in Massachusetts have been in contact with at least 10 other state laboratories interested in adding the TREC assay and SCID to their screening panel. Wisconsin will begin working with Louisiana to initiate screening for SCID sometime after July 1, 2010. Currently, Wisconsin and Massachusetts are the only two states that screen for SCID.

“State public health laboratories and newborn screening lead the way in identifying infants with life threatening conditions. This has always been part of our public health mission, and we believe that there will be additional opportunities to expand newborn screening as a result of the work here in Wisconsin,” says Dr. Charles D. Brokopp, Director of the Wisconsin State Laboratory of Hygiene.

According to preliminary data from the Children's Hospital of Wisconsin, a single baby with a late SCID diagnosis costs an average of \$2.2 million. Medical care for one baby with an early SCID diagnosis costs \$250,000. Testing the 70,000 babies born annually in Wisconsin for SCID as part of the routine newborn screening panel costs approximately \$350,000 (\$4-5 per test). According to Dr. Brokopp, “The savings from one positive diagnosis pays for testing of all babies for the entire year.”

The Wisconsin State Laboratory of Hygiene is a member of the Association of Public Health Laboratories (APHL). APHL's Newborn Screening and Genetics Program works to strengthen the role of public health laboratories in genetic testing and designs strategies to address changes in the newborn screening testing field. The association collaborates with the Centers for Disease Control and Prevention to provide input on quality control and proficiency testing issues relevant to newborn screening laboratories across the globe.

The Association of Public Health Laboratories is a national non-profit located in Silver Spring, MD, that is dedicated to working with members to strengthen governmental laboratories with a public health mandate. By promoting effective programs and public policy, APHL strives to provide public health laboratories with the resources and infrastructure needed to protect the health of US residents and to prevent and control disease globally.

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