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EMBARGOED RELEASE:
JANUARY 16

Jeffrey Modell Foundation, Wisconsin State Laboratory Of Hygiene And Children's Hospital Of Wisconsin Launch Pilot Newborn Screening Program To Detect and Treat "Bubble Boy Disease"

MILWAUKEE (1/12/06) - Health researchers in Wisconsin today announced a first of its kind newborn screening study aimed at detecting a rare but often undiagnosed immune system disease that is fatal without treatment and believed by some doctors to be responsible for a number of unexplained infant deaths.

The pilot program will be a collaborative effort between the State Laboratory of Hygiene at UW-Madison and Children's Hospital of Wisconsin in Milwaukee. Using residual blood specimens from Wisconsin's nationally recognized newborn screening program, the pilot program will develop the protocols to screen all newborns for Severe Combined Immune Deficiency disease (SCID), sometimes known as "Bubble Boy Disease." The goal is to extend routine screening for SCID to every newborn in Wisconsin, estimated at 70,000 annually.

"SCID can now be cured with a relatively simple bone marrow transplant, if diagnosed in the first weeks or months of life. We believe routine screening of all newborns will find more SCID babies whose disease in the past may have been masqueraded as unexplained deaths in early infancy. If their problem had been identified correctly, early on, and treated properly through a screening program, those babies might be alive today," said John Routes, MD, medical director of Allergy and Clinical Immunology at Children's Hospital of Wisconsin.

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"Today's highly sophisticated Newborn Screening Programs, such as Wisconsin's, where we routinely screen for 47 parameters, as well as the technology to screen for SCID, suggest that the time has come to add this capability to our country's highly effective programs. We welcome the opportunity to partner with the Modell Foundation and Children's Hospital to undertake the next important first step," said Ronald H. Laessig, PhD, Emeritus Director and Professor of Population Health Sciences at the State Laboratory of Hygiene, UW-Madison.

Working quietly behind the scenes, the Jeffrey Modell Foundation (JMF), a world authority on primary immunodeficiency diseases, has spent months gathering scientists and public health leaders together to help drive the SCID screening initiative. Several other states also are actively considering SCID screening programs. JMF and Children's Hospital are providing the funding for this pilot.

"SCID is a pediatric medical emergency nationwide. But we now can diagnose SCID with 99 percent accuracy. Once we identify these infants, we can treat them with a 95 percent success rate using bone marrow transplantation. If we catch it early with a simple blood test, we can cure a baby with SCID. If we wait, we will spend millions of dollars on long, unneeded hospitalizations that are destined to have a grim outcome," said Fred Modell, of New York City, a founder of JMF, along with his wife Vicki.

JMF, using its own funds and a dedicated network of supporters, successfully has rallied leaders in the federal and state governments, pediatric medicine and pharmaceutical industry behind its cause. Several other states are lined up to follow Wisconsin's lead. They include California, Maryland, Massachusetts, Missouri and New York. Each is now considering similar pilot programs. State public health officials convened in Atlanta in November at a workshop addressing newborn screening for SCID. The meeting was convened by the U.S. Centers for Disease Control and Prevention (CDC) and funded by JMF.

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"With 70,000 births a year in Wisconsin, the data suggests that in 2007 we will save at least one baby's life in that state alone, thanks to the leadership role public officials are taking there. We need this pilot program to demonstrate the feasibility of routine testing and ultimately early treatment. We will need mo re federal and state partners and more support from across the country if we are going to save more lives," said Vicki Modell.

The Modells lost their son Jeffrey at the age of 15 when he died of complications from a primary immunodeficiency disease. There are more than 140 different primary immunodeficiency diseases caused by an immune system that does not function properly. The genetically passed on condition affects as many as 1 million Americans, mostly children.

"The Boy in the Bubble"

SCID is the most lethal version of all Primary Immunodeficiency diseases. It is often called "Boy in Bubble" disease after the movie of the same name which starred John Travolta, the true story of a boy with SCID who died at age 12 after spending his life in a plastic bubble because he was so vulnerable to infection.

SCID causes a defect in the white blood cells that help protect the body from viruses, bacteria and fungi. Doctors know that SCID is the result of a mutation in one of at least 12 genes, and bone marrow transplants are presently the best treatment.

"Fred and Vicki Modell have dedicated their lives to raising awareness and improving diagnosis and treatment of primary immunodeficiency diseases such as SCID. They have made the JMF an effective, efficient force in overcoming numerous obstacles that until now blocked implementation of a screening program that could detect this fatal disease," added Dr. Routes.

Obstacles Overcome

Because of research and dialogue promoted by the Modells and the Centers for Disease Control and Prevention, advocates for SCID screening have built a groundswell of support and began to resolve issues involving parental consent, institutional review board concerns, variability in assays in the blood testing used for screening, cost concerns and understanding about the availability of new treatment.

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The primary investigators, Dr. Routes and Dr. Laessig are developing the laboratory testing protocol to demonstrate the feasibility of routine screening. Children's Hospital of Wisconsin will take the lead in diagnosis and treatment of potential SCID babies. Once a workable program is demonstrated, the investigators foresee a rapid acceptance of routine screening and treatment protocols.

CDC Comments

"The workshop held last November generated results far beyond anyone's expectations. The Jeffrey Modell Foundation identified newborn screening of SCID as a primary focus of its efforts and has now created a unique federal-state-private collaboration. We are committed to their vision that all newborns will be screened for SCID in a system where true cases are identified promptly and treated effectively," said Dr. Robert Vogt of the Newborn Screening Branch of the CDC Division of Laboratory Sciences.

SCID Babies Look Normal at First

"When these babies are first born, they look perfectly normal. They're fine until they get sick with very bad infections. Once the baby starts developing severe infections and spends lots of time in the hospital, medical bills can top $1 million," said Rebecca Buckley, MD, a professor of pediatrics and immunology at Duke University School of Medicine, and an advocate with the Modells for national screening for SCID who spoke at the CDC workshop.

She said babies usually receive a bone marrow transplant from a parent or sibling who can provide stem cells that can develop into healthy blood cells in the baby. The baby receives the cells slowly through a syringe. She has watched some of the babies she has treated grow up to graduate from college, go to medical school or graduate from business school.

"I used to see these babies die all the time. Now it's just wonderful to be able to see them make it and grow up to be normal," she said.

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About Children's Hospital of Wisconsin

Children's Hospital of Wisconsin, founded in 1894, is recognized as one of the leading pediatric health care centers in the United States and is on Child magazine's 10 Best Children's Hospitals list. Children's Hospital is the only freestanding Level I Trauma Center in the region that cares exclusively for children and adolescents. The hospital has been designated a Magnet hospital by the American Nurses Credentialing Center, a national honor that recognizes nursing excellence. Children's Hospital is the flagship member of Children's Hospital and Health System. Private, independent and not-for-profit, the hospital serves children and families from Wisconsin, the Upper Peninsula of Michigan, northern Illinois and beyond.

About Jeffrey Modell Foundation

The Jeffrey Modell Foundation (JMF) was established in 1987 by Vicki and Fred Modell in memory of their son Jeffrey, who died at the age of 15 of a PI disease. The Foundation is dedicated to early and precise diagnosis, meaningful treatments, and ultimately cures of Primary Immunodeficiencies. Today there are 32 Jeffrey Modell Research and Diagnostic Centers and 120 Referral Centers worldwide. Construction has begun for the Jeffrey Modell Immunology Center on the main quad of the Harvard Medical School. More information about PI can be found at www.info4pi.org or by calling the JMF hotline at 1-866-INFO-4-PI.

About the Wisconsin State Laboratory of Hygiene

The State Laboratory of Hygiene is Wisconsin's Public Health Laboratory. It is a nationally and internationally recognized leader in the field of Newborn Screening. As part of the University of Wisconsin-Madison School of Medicine and Public Health, it has played an instrumental role in developing many innovations now routine in newborn screening programs. The State Laboratory was the first to develop and fully implement routine screening for cystic fibrosis coupled with a vigorous and effective "pre-crisis" treatment program. Children identified with a "positive" CF test are now routinely treated at children's hospitals in Milwaukee and Madison. Currently, 35 states have followed the Wisconsin lead and now screen for CF.

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