Wisconsin State Laboratory of Hygiene
UNIVERSITY OF WISCONSIN–MADISON

Information about the Wisconsin Newborn Screening Pompe Pilot Project

Project title:
Evaluation of a Newborn Screening Process that Facilitates Early Identification and Treatment of Infants with Pompe Disease

To request additional information:
Call 1-844-247-3255 to leave a message or send a message to Pompe@slh.wisc.edu

This sheet provides a brief description of Pompe disease and where additional information can be found.

Newborn screening is a standard public health care service that screens newborns for a variety of treatable health problems they may be born with. The program identifies babies who may have these "congenital conditions," and provides referrals as needed for follow-up confirmatory testing, clinical care, and family counseling. Newborn screening is performed throughout the United States and in all developed countries around the world.

Wisconsin is studying a new screening process for a disorder called Pompe disease, which was recommended to be included in newborn screening programs by a committee at the U.S. Department of Health and Human Services, and officially added to the Recommended Uniform Newborn Screening Panel on March 2, 2015. Other states are already screening or planning to start screening for Pompe disease.

The Wisconsin study is funded by the National Institutes of Health to evaluate a process for screening for Pompe disease in newborns for a period of 12 months. The experience gained from this study will help us decide whether this way of screening for Pompe disease should become a part of routine newborn screening.

Pompe disease is a rare disorder, affecting about one in every 40,000 newborns. Pompe disease causes damage to certain organs and tissues, especially the heart and the muscles used for breathing. How bad the disease is can vary widely, but some affected infants will die in the first year of life if not treated. This is called the "classic infantile" form of the disease. In the "late onset" form of the disease, individuals may not become ill until later in childhood, teenage years, or older.

Newborn screening for Pompe disease allows for early disease detection and treatment, which have been proven to lead to better health outcomes for many children with the infantile form and may even be life-saving. This study uses the same blood already collected by heel-stick for standard newborn screening currently done for every baby born in the state of Wisconsin.

If a baby's screening test result is positive for Pompe disease, heart function is promptly checked and additional blood is drawn to see if the baby really does have Pompe disease. Confirmatory testing and treatment will be overseen by specialists at Children's Hospital of Wisconsin/Medical College of Wisconsin, or University of Wisconsin American Family Children's Hospital.

The benefit of taking part in this study is early detection and treatment of children with the infantile form of Pompe disease.

A risk of the study is that the screening process may identify children with a less severe form of Pompe disease, called "late-onset disease." People with "late-onset disease" may not have symptoms until later in childhood or in adult life. This could lead parents, children, or adults to worry a great deal waiting for the illness to strike. However, this earlier knowledge of the late-onset form of Pompe disease could also be a benefit, allowing for earlier diagnosis and treatment later in life since people with late-onset Pompe disease often have symptoms for years before their physicians make a correct diagnosis of Pompe disease.

The screening test could also lead to a false positive result – that is, a child who is completely healthy and will never be affected with Pompe disease can have an abnormal screening test. Doctors can find out which children actually have the disease and which do not during the confirmatory process; a false positive result can usually be identified within two weeks.

To participate in this study parents don't have to do anything, since all babies will be tested unless a parent decides they would prefer not to be a part of the study. No extra blood is required to do the test, and being in this study will not cost parents or their insurance company any extra money. The baby's doctor or specialist will call the parents if the baby has a positive (abnormal) screening result.

Here is a link to more information about Pompe disease: http://ghr.nlm.nih.gov/condition/pompe-disease

Here is a link to more information about this study: http://www.slh.wisc.edu/clinical/newborn/pompe-pilot-project/

If you want to talk to an expert about this study, leave a message with your phone number at 1-844-247-3255 and we will call you back.

If you do NOT want your newborn child to be part of this study,call 1-844-247-3255 and leave a message stating:

- Mother's full name
- Due date
- Anticipated or known birth facility, or specify an out of hospital birth
- That you decline participation in this Pompe newborn screening project
- Phone number to allow us to call and confirm status.