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Information about the Newborn Screening X-Linked Adrenoleukodystrophy Demonstration Project

Project Title: Evaluation of a Newborn Screening Process that Facilitates Early Identification and Treatment of Infants with X-Linked Adrenoleukodystrophy

To opt out or request additional information: Call 1-833-828-2803 to leave a message
Send a message to NbsOptOut@slh.wisc.edu

This sheet provides a brief description of X-Linked Adrenoleukodystrophy (X-ALD) and information about where you can get additional information, including contacts for questions.

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

Wisconsin is evaluating a new screening process for X-ALD, a condition that was recently recommended for inclusion in newborn screening programs by the US Secretary of Health and Human Services. Other states are already screening or planning to start screening for X-ALD. In Wisconsin, on May 12, 2022, the Secretary of the Wisconsin Department of Health Services approved the addition of X-ALD to the newborn screening panel following the administrative rule-making process. The rule-making process is ongoing and X-ALD is not yet approved for formal addition to the Wisconsin Newborn Screening Program.

The Wisconsin demonstration project is funded by the US Centers for Disease Control and Prevention to evaluate a process for screening for X-ALD in newborns for a period of 18 months. The experience gained from this project should be helpful when the current planned process of screening for X-ALD is anticipated to become a part of routine newborn screening.

X-ALD is a rare disorder caused by a change in a gene that makes a protein that helps the body break down certain types of fats. This disease affects both males and females, but females tend not to develop symptoms until adulthood. Males with X-ALD are often normal in infancy, but they may go on to develop problems with their adrenal glands, brain, and spinal cord. Treatments for X-ALD include cortisol replacment for adrenal dysfunction and hematopoietic stem cell transplantation (HSCT) to arrest progressive brain abnormalities. Recently, a new gene therapy was approved for individuals 4-17 years old. If untreated, about 40% of affacted males will develop rapidly progressive brain damage in childhood; this is called childhood cerebral ALD (CCALD). CCALD leads to cognitive loss, blindness, severe disability, and death. CCALD can be treated with HSCT or gene therapy, but treatment is generally only effective when CCALD is identified at an early stage, when brain changes are seen on brain imaging studies but before the development of clinical symptoms. Without treatment, these boys may become seriously ill or develop irreversible neurologic injury during childhood. People with "late onset disease" may not have symptoms until later in childhood or in adult life.

Newborn screening for X-ALD allows early disease detection and treatment, which could lead to better outcomes for children with the CCALD form. This project uses the same blood already collected by heel-stick for standard newborn screening.

If a baby's screening test result is positive for X-ALD, additional blood is drawn to see if the baby really does have X-ALD. Confirmatory testing and treatment will be overseen by specialists at University of Wisconsin American Family Children's Hospital or Children's Wisconsin/Medical College of Wisconsin. The screening test could lead to a false positive result - that is, a child who is healthy and will never be affected with X-ALD might have a screening positive result. Doctors can find out which children have the disease and which do not during the confirmatory process; false positive findings can usually be identified within two to four weeks.

To participate, you don't have to do anything, because all babies will be tested shortly after birth unless a parent decides they would prefer their child not participate. No extra blood is required to do the test and participating will not cost 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 X-ALD: https://www.babysfirsttest.org/newborn-screening/conditions/adrenoleukodystrophy

If you want to talk to an expert about this project, leave a message including your phone number at 1-833-828-2803, and we will call you back as needed.

If you do NOT want your newborn child to be part of this study, call 1-833-828-2803, and leave a message stating:

Mothers full name; due date; anticipated or known birth facility, or noting out of hospital birth; that you decline participation in the X-ALD demonstration project; and a phone number to allow us to call and confirm status.

Thank you for your consideration.

Sincerely,

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