FIREFLY FUND COMMITS $500K TO NOVEL NEWBORN SCREENING STUDY THAT WILL HELP IDENTIFY RARE DISEASES AT BIRTH

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Austin Nonprofit Commits $500K to Novel Newborn Screening Study That Will Help Identify Rare Diseases at Birth

Pilot Study enrolls first neonate, first time anywhere in the world a newborn is screened for Niemann-Pick Disease Type C at birth

AUSTIN, TX – Firefly Fund, a nonprofit focused on finding a cure for Niemann-Pick Disease Type C (NPC), a rare genetic neurodegenerative disease, announced today its five-year $500,000 commitment to the largest multi-disorder, consented pilot newborn screening study in the country. Earlier today at Montefiore Hospital in New York, with a quick heel prick and a smudgy blood spot on a newborn screening card, the first neonate was enrolled into the study.

The pilot study, ScreenPlus, is led by chief of Pediatric Genetic Medicine Dr. Melissa Wasserstein at the Children's Hospital at Montefiore, and is the first time any newborn, anywhere in the world is being screened for NPC. New parents are given the option to participate in the study on top of the routine newborn screening conducted by the New York State Department of Health. NPC is one of about a dozen rare diseases on the ScreenPlus panel. The study aims to screen about 175,000 ethnically-diverse babies born at one of 8 hospitals across New York. Newborn screening is a public health program and has been an integral part of preventing rare diseases in children and babies for over five decades.

"A report from the 2020 census found that there were 3.6 million births in the US last year. That means that as many as 36 families gave birth to a beautiful baby who might also – completely unbeknownst to them – have NPC. ScreenPlus is a chance to reverse this at long last," said Pam Andrews, co-founder and executive director of Firefly Fund. "Knowing sooner is a chance to intervene sooner and help these babies live longer and healthier lives."

Firefly Fund was founded by Pam and Chris Andrews after their daughters Belle and Abby were diagnosed with NPC at the ages of 5 and 2, respectively. Belle was diagnosed following a long diagnostic odyssey and significant onset of symptoms. Abby, who was tested for the genetic disease following her older sister's diagnosis, was able to be diagnosed and treated before the onset of symptoms.

"More and more the science is telling us that the earlier we can intervene in this devastating disease, the better chance we have of improving patient outcomes. But first, we have to identify the children," Andrews said. "We are so grateful to Firefly Fund donors and the entire NPC Community for the continued support, which allows us and inspires us to do this important work."

In addition to Firefly Fund's commitment, the National Institutes of Health awarded Wasserstein $3.2 million to conduct ScreenPlus. The program is also funded through a unique cost-sharing collaboration between among, several leading pharmaceutical companies and rare disease foundations focused on treating rare diseases.

Any infant who tests positive for NPC through the study will be referred for specialized care and will undergo long term follow up. The study's findings will determine whether NPC and any of the other diseases on the ScreenPlus panel should be added to routine newborn screening panels nationwide.

"By identifying babies with complex disorders like NPC as early as possible, we have the opportunity to provide FDA-approved treatments or connect them to clinical trials," Wasserstein said. "We believe ScreenPlus will add to the scientific body of knowledge about newborn screening but also, importantly, early diagnosis will allow parents to connect with expert clinicians and consider intervention before life-threatening symptoms take hold of their child."

NPC is a rare genetic neurodegenerative disease that currently has no cure. It is progressive with worsening symptoms that include enlargement of the liver and spleen, slurred speech, lack of muscle coordination, and tremors or difficulty with fine motor skill development. The disease typically begins in early childhood and worsens over time. NPC expert clinicians agree that early diagnosis and intervention, before the onset of visible signs or symptoms of the disease, improves health outcomes.

In 2017, with the support of the NPC Community, Firefly Fund convened a multi-disciplinary working group comprised of NPC community and industry stakeholders. This Working Group of clinicians, scientists, researchers, government officials, and advocates is Co-Chaired by Dr. Elizabeth Berry-Kravis of Rush University Medical Center and Dr. Marc Patterson of Mayo Clinic.

About Firefly Fund

Firefly Fund is a 501(c)3 nonprofit organization founded in March 2017 to fund the research and education necessary to accelerate a cure for rare neurodegenerative genetic diseases that affect children and currently have no cure—starting with Niemann-Pick Disease Type C (NPC). Headquartered in Austin, Texas, Firefly Fund leads a collection of programs including Newborn Screening, Translational Medical Research, and a Patient Access Fund. These programs offer resources, support, and reassurance to NPC families. A rare disease diagnosis is unimaginable, but together we will find a cure. For more information, visit www.fireflyfund.org.