Children’s Mercy Kansas City, University of Utah & RI-MUHC Receive $3.1 Million NIH Grant to Study Rare Genetic Disease

KANSAS CITY, MO, UNITED STATES, September 25, 2024 /EINPresswire.com/ -- In a significant advancement for pediatric medicine, an international team of researchers from Children’s Mercy Kansas City, the University of Utah, and the Research Institute of the McGill University Health Centre (RI-MUCH) in Montreal, Canada have been awarded a $3.1 million grant by the National Institutes of Health (NIH) to support groundbreaking research in Leukodystrophy, a rare genetic disorder that affects 1 in 5,000 children causing progressive loss of neurological function.

More than 50 types of Leukodystrophy have been identified, which can cause a range of symptoms including seizures and intellectual impairment. Swift diagnosis and intervention are critical as most children diagnosed with Leukodystrophy die before their teenage years.

The research aims to address the diagnostic challenges by leveraging advanced genome sequencing technologies spearheaded by the Genomic Answers for Kids program (GA4K) at Children’s Mercy.

“We are using long-read genome sequencing (HiFi-GS) to uncover genetic variants that are not detectable with standard short-read sequencing to increase the diagnostic rate for complex cases,” said Tomi Pastinen, MD, PhD., Director, Genomic Medicine Center, Children’s Mercy and the Contract Principal Investigator (PI) of the study. “Collaborating with the other PIs, Dr. Josh Bonkowsky from the University of Utah and Dr. Geneviève Bernard from the RI-MUHC on this research underscores our global commitment to addressing rare disease in hopes of accelerating the delivery of crucial interventions and providing answers to families sooner.”

The urgency of this research is underscored by the story of Conner, diagnosed with cerebral adrenoleukodystrophy at just four years old.

“Without immediate treatment, he would have lost his ability to see, hear, eat, walk and communicate - falling into a vegetative state before dying,” said Kirsten Finn, Conner’s mother. "The research funded by this grant is not just important—it's a lifeline. It offers hope to families like ours that one day, this narrow treatment window will be a thing of the past."

Dr. Pastinen will take the stage at this year’s RARE Advocacy Summit to share more on this research along with other GA4K updates, “Genomic Answers for Kids is leading the way in

diagnosing kids with rare disease with over 8,000 children and total of 15,300 family members involved. We are excited to share this is the fourth NIH grant supporting the program to date," Dr. Pastinen said.

About Genomic Answers for Kids

Genomic Answers for kids is a Children's Mercy research initiative to build a first-of-its-kind pediatric data repository to facilitate the search for answers and novel treatments for pediatric genetic conditions. The goal is to collect genomic data and health information for 30,000 children and their families over seven years, creating a database of nearly 100,000 genomes.

Learn more about Genomic Answers for Kids.

About Children's Mercy Kansas City

Founded in 1897, Children's Mercy is a leading independent children's health organization dedicated to holistic care, translational research, educating caregivers and breakthrough innovation to create a world of well-being for all children. With not-for-profit hospitals in Missouri and Kansas, and numerous specialty clinics in both states, Children's Mercy provides the highest level of care for children from birth through the age of 21. U.S. News & World Report has repeatedly ranked Children's Mercy as one of "America's Best Children's Hospitals." For the fifth consecutive time in a row, Children's Mercy has achieved Magnet nursing designation, awarded to only about 8% of all hospitals nationally, for excellence in quality care. More than 800 pediatric specialists, researchers and faculty are actively involved in clinical care, pediatric research and education of the next generation of pediatric specialists. Thanks to generous philanthropic and volunteer support, Children's Mercy provides hope, comfort and the prospect of brighter tomorrows to every child who passes through its doors. Visit Children's Mercy and the Children's Mercy Research Institute to learn more, and follow us on Facebook, LinkedIn, Twitter, Instagram and YouTube for the latest news and videos.

About RARE Advocacy Summit

For the first time, Global Genes and RareKC are coming together to host the RARE Advocacy Summit Sept. 26-27 in Kansas City. Global Genes is taking the Summit on the road to Kansas City, after having hosted this annual event in California for over 12 years. The Rare Advocacy Summit is one of the world's largest gatherings of rare disease patients, caregivers, advocates, healthcare professionals, researchers, partners, and allies at the RARE Advocacy Summit to work together to build a path to hope. View the full conference agenda.

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