The Newborn Screening Ontario Program Begins Screening for GAMT

CARLSBAD, CA, USA, October 29, 2022 /EINPresswire.com/ -- Guanidinoacetate methyltransferase (GAMT) deficiency is now one of the disorders that lab technicians are looking for newborns in Ontario, Canada. The program began screening for GAMT deficiency on Monday, Oct. 17. GAMT is an inherited disorder that primarily affects the brain and muscles.

This addition is due in large part to the advocacy of Association for Creatine Deficiencies' Scientific Medical Advisory Board member, Dr. Andreas Schulze. Dr. Schulze has worked at the Hospital for Sick Children in Toronto, Canada, since 2007 and has established his own research group with a focus on Cerebral Creatine Deficiency Syndromes, Regulation of Creatine Synthesis, Pathophysiology of Guanidino Compounds, and Small Molecule Treatments. He is the director of the newborn screening program at the Hospital for Sick Children. Dr. Schulze was the first to report and describe the full biochemical spectrum in GAMT Deficiency and has an ongoing interest in advancing research of creatine deficiencies.

"Thank you to all of those that supported Dr. Schulze's nomination to make this happen. A diagnosis of GAMT at birth brings the chance of a full life, free of disabilities for those with GAMT. We are grateful for Dr. Schulze's commitment to GAMT," said Heidi Wallis, ACD executive director.

GAMT deficiency is an inherited condition that affects the body's ability to produce creatine. Without an adequate supply of creatine, the body is unable to use and store energy properly. This can cause developmental delay, speech problems, seizures and behavior issues. Lack of early treatment can lead to lifelong cognitive impairments which can be severe. Starting the dietary and medical treatment early in life before symptoms arise is most effective.

"Tenacity prevails! I am so happy for the fact that children with GAMT born in the province of Ontario will be identified early on and receive their treatments in time from now on," said Dr. Schulze. "With that they can grow up healthy and happy. The accomplishment of having GAMT added to the screening panel has many parents. I am more than grateful to our families for their support, to my colleagues here and abroad that were involved, to the whole team of Newborn Screening Ontario, and to the members of the advisory committee. I cannot wait to counsel and guide the first GAMT family identified by our newborn screening."

ACD is committed to providing patient, family and public education to advocate for early

intervention through newborn screening, and to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes.

Wallis personally knows the importance of early diagnosis and treatment. “I have two children with GAMT, and the difference a diagnosis at birth makes is nothing short of life-changing,” Wallis said. “My daughter, who was diagnosed at 5 years old, has recurrent seizures that cause her to sustain serious injuries. She is intellectually disabled and will need constant care the rest of her life. My son, diagnosed and treated at birth, is a neurotypical 10-year-old and will no doubt become an independent and contributing member of society.”

In May, ACD announced that after six years of advocacy efforts, the United States Advisory Committee on Heritable Disorders in Newborns and Children voted unanimously to advise the Secretary of Health and Human Services Xavier Becerra to add GAMT to the Recommended Uniform Screening Panel (RUSP). The RUSP provides state newborn screening programs with a carefully curated list of disorders that meet the committee’s criteria for inclusion. GAMT is expected to be added to the RUSP upon HHS Secretary Becerra’s approval in November.

About ACD: The Association for Creatine Deficiencies’ mission is to eliminate the challenges of CCDS. ACD is committed to providing patient, family, and public education to advocate for early intervention through newborn screening, and to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes. Because CCDS mimic symptoms of other medical conditions, patients are often first diagnosed with autism, cerebral palsy, epilepsy, and other disorders. Proper diagnosis and early intervention are critical to establishing interventions needed to improve life quality and longevity for the CCDS patient. For more information regarding ACD, please visit creatineinfo.org.

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