First Baby with GAMT Deficiency Identified through Newborn Screening in Australia

CARLSBAD, CA, UNITED STATES, June 29, 2023 /EINPresswire.com/ -- Today the Association for Creatine Deficiencies (ACD) announced that a baby has been identified through newborn screening with Guanidinoacetate Methyltransferase (GAMT) Deficiency in Victoria, Australia. The Victoria newborn screening program began screening for GAMT, under the supervision of Dr. James Pitt, head of biochemical genetics for the Victorian Clinical Genetics Services at the Royal Children's Hospital in Victoria, in the early 2000s when the lab first began using mass spectrometry in their routine newborn screenings.

The baby identified by Pitt's lab in 2022 is a healthy baby girl from Melbourne, and her parents report being overwhelmed with gratitude. "We were shocked to learn that our daughter had GAMT. I'm sure most parents would agree the thought of anything being wrong with your baby hits home very hard and isn't something you expect," said Josh Cook. "Like the majority of Australians, we had never heard of this condition. After learning of the devastating impact GAMT has on an individual's quality of life if not detected early we can hardly express how truly grateful we are to Dr. James Pitt and the Victorian lab for their continued commitment to screen for GAMT. We will be forever grateful for this program and want to pay it forward and see all other Australian states and other countries include GAMT in their newborn screening."

"Our daughter is developing great so far, as we have been reassured by doctors. She began taking supplements when she was about seven weeks old and it quickly became a part of our daily routine. It is almost too difficult to imagine the life she was going to lead, with irreparable brain damage, if it weren't for newborn screening," says Amy Cook. "My wish is to see GAMT included in newborn screening worldwide so that a child's quality of life is not based simply on the location they were born in."

ACD Executive Director Heidi Wallis commented, “I have a daughter who was diagnosed with GAMT at five years old, after irreparable brain damage had occurred. Starting creatine at that age helped improve some of her symptoms, but she is 19 now and suffers from epilepsy and intellectual disability. My youngest child was diagnosed at birth thanks to his big sister. He is a typical 11-year-old boy today who loves computer games, sports, and reading, and he has never had a seizure or required the therapies my daughter has needed. When a child is diagnosed after symptoms appear, the family is forever impacted and society is as well. The family of this baby was so fortunate to be born in Victoria. Victoria is one of only a handful of newborn screening programs in the world that currently include GAMT in their screenings. ACD is working on changing this.”

Newborn screening is a routine health screening of infants that is conducted by most industrialized countries around the globe. It is sometimes referred to as the “PKU test” or “heel prick test” after its method, a heel prick to collect the blood, and the first newborn screen that was launched in the 1960s to detect Phenylketonuria, or PKU. Newborn screening programs screen for as many as 50 disorders in some locations, but this is not consistent. In Australia, the only state screening for GAMT is Victoria. In the United States, only Utah, New York, and Michigan currently screen for GAMT. The future of a child should not be determined by a family’s address. GAMT is not obvious when a baby is born. For instance, there are no dysmorphic features or clear, immediate symptoms to know that creatine supplements need to begin right away. “Without newborn screening, there is very little hope of an early diagnosis,” states Wallis. “With newborn screening, a person with GAMT may live a typical, healthy life, with very easily accessed and affordable supplements.”

ACD has been advocating for increased GAMT screening globally since its formation in 2012 and GAMT was added to the United States’ Recommended Uniform Screening Panel in January of this year. To learn more about GAMT newborn screening, please contact ACD. The organization aims to help those who want to become advocates and present data to those in newborn screening programs that would like to enhance their screenings by adding this condition.

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 http://www.creatineinfo.org.

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