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FIRST BABY WITH GAMT DEFICIENCY IDENTIFIED THROUGH NEWBORN SCREENING

Carlsbad, CA (Dec. 30, 2020)—For the first time, a newborn baby has been diagnosed with Guanidinoacetate methyltransferase (GAMT) deficiency through newborn screening. The baby boy was identified through the state of Utah's newborn screening program, which has tested all babies born in Utah for GAMT since 2015. This is the first newborn identified with GAMT solely by newborn screening and without a family history. This infant's diagnosis will likely open the door to many more states and countries adding GAMT to their screening panels.

GAMT Deficiency is caused by mutations in the GAMT gene. Due to a lack of the GAMT enzyme, patients with GAMT are unable to break down the guanidinoacetate (GAA) formed in the first step of creatine synthesis. This results in a buildup of GAA which is believed to be toxic to the brain at high levels and causes damage that accumulates over time. With treatment early in life, severe neurological symptoms including intellectual disability, limited speech development, recurrent seizures, autistic-like behavior and involuntary movements can all be avoided. Treatment is aimed at reducing GAA production and supplying the creatine that is not produced by the body. Patients are typically prescribed oral supplements of creatine monohydrate and l-ornithine. Some physicians also recommend diet restrictions and sodium benzoate supplementation to further minimize GAA accumulation.

The newly diagnosed infant, Woodward Tribe, will be able to live a typical, healthy life because he can receive treatment for GAMT, an otherwise debilitating disorder, at such a young age, thanks to the newborn screening test. His parents, Stewart and Becky Tribe, shared their experience with their son's diagnosis, and according to Stewart Tribe, "During the COVID lock-down in April, we decided to make the move from California to Utah. It was an impulsive decision at the time that we now recognize as nothing short of an alignment of stars for our family and newborn son Woodward.

"Had we not moved and delivered Woodward in a hospital in Utah, he never would have been screened for GAMT," Tribe continued. "Without Utah's newborn screening we'd likely spend years frantically searching for a cause to Woodward's developmental delays. But thanks to a community of highly specialized doctors, dietitians and passionate parents with children with GAMT—who all just happen to be located here in Utah—we don't have to. We're stunned by the serendipity of it all and beyond-words grateful for the treatment Woodward is now receiving at such a young and crucial age."

Advocacy for the inclusion of GAMT on newborn screening panels has been a major ongoing focus for the ACD. The organization's Newborn Screening Committee has advocated at both the federal and state levels across the United States. The ACD is committed to continuing advocacy efforts for inclusion of GAMT on the federal Recommended Uniform Screening Panel (RUSP). GAMT was first nominated for review by the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC) in 2016 by Dr. Nicola Longo, Professor of Pediatrics and Chief of the Division of Medical Genetics at the University of Utah.



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The ACHDNC advises the Secretary of Health and Human Services on adding new disorders to the RUSP. The Secretary of Health makes the final decision on which disorders are added to the RUSP and many states will not consider adding a disorder to their panel until it is added to the RUSP. When GAMT was initially nominated in 2016, the ACHDNC did not recommend GAMT for additional consideration due to the fact that an infant had not yet been identified by newborn screening as having GAMT.

"Thanks to this successful screening, the outlook is bright for infants born with GAMT in the future," says Heidi Wallis, a parent of two children with GAMT and President of the ACD. "The ACD plans to return to the ACHDNC as soon as possible to ask for inclusion of GAMT on the RUSP. From there we will widen our advocacy work to focus on all countries with a newborn screening program."

Utah began screening for GAMT June 1, 2015 and has screened approximately 275,000 babies in that time. Until more infants are identified through newborn screening, the true incident rate is unknown.

"GAMT deficiency is very rare and difficult to diagnose clinically since developmental delays and seizures have a multitude of causes. It can be treated with nutritional supplements that are widely available and relatively inexpensive. Therapy needs to be started early in life to prevent brain damage. The first case identified by newborn screening worldwide demonstrates that GAMT deficiency can be detected at birth before any brain damage has occurred," said Dr. Longo.

According to Marzia Pasquali, Professor of Pathology and Section Chief of Biochemical Genetics at ARUP Laboratories at the University of Utah, "It is only fitting that the first case is being reported in Utah since this state was the first in the USA to develop a screening method and the first to initiate screening for GAMT deficiency in 2015. Initial testing on 10,000 de-identified blood spots was able to correctly detect those from patients with GAMT deficiency, with no false positive or false negative results. Screening for GAMT deficiency can easily be integrated to the standard newborn screening using tandem mass spectrometry, with a minimal incremental cost. We hope that the identification of this case will lead to the inclusion of GAMT deficiency in newborn screening programs worldwide."

Utah Newborn Screening Program Manager Kim Hart emphasized the significance of this major milestone, saying, "GAMT is a poster child for newborn screening - it is inexpensive to screen for and has an effective treatment. Early diagnosis and treatment truly changes lives for these families."

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 screening and treatments 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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