ASSOCIATION FOR CREATINE DEFICIENCIES

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FOR IMMEDIATE RELEASE

GAMT Deficiency Advances Towards Universal Newborn Screening

Carlsbad, CA (August 12, 2021)—The United States Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) voted unanimously today to advance Guanidinoacetate Methyltransferase Deficiency (GAMT) forward for review by the Evidence Review Committee to add GAMT to the Recommended Uniform Screening Panel for all newborn babies. The Evidence Review Committee may take as long as nine months to review the condition for feasibility of newborn screening in state laboratory programs. When the review is completed, the Evidence Review Committee will recommend whether to advance GAMT Deficiency to the Secretary of Health and Human Services Xavier Becerra, for final review and addition to the United States' Recommended Uniform Screening Panel (RUSP). The RUSP is a tool used by most states in establishing which disorders they will include in their newborn screening panel. Additionally, many states have recently adopted legislation that holds their laboratories accountable to add new conditions added to the RUSP in a timely manner.

"With newborn screening for GAMT, patients will be identified before debilitating long-term effects begin. Families will be able to manage this condition from birth and help their child reach their full potential. GAMT is a treatable condition, discovered 27 years ago, and this vote marks the beginning of the end of suffering for families like ours. Future families will no longer have to witness the decline of their child and search for answers on their own," states ACD President Heidi Wallis.

The ACHDNC meets quarterly to discuss disorders that are serious if not diagnosed at birth and have a suitable treatment and newborn screening method. This is the second time GAMT has been officially nominated for consideration by the ACHDNC. In 2016, GAMT was nominated by Dr. Nicola Longo and was reviewed by the ACHDNC. GAMT was not moved forward to evidence review at that time because an infant had not been diagnosed with GAMT through the use of prospective newborn screening. Thanks to the advocacy work of parents through ACD, as well as the tireless dedication of many researchers and scientists, GAMT was added to the Utah and New York screening panels. Two infants recently screened positive for GAMT, allowing a renomination of the disorder and today's critical step forward towards universal GAMT newborn screening in the United States.

"We look forward to the findings of the Evidence Review Committee and advocating at the state level to promote the adoption of GAMT on all newborn screening panels," commented Kim Tuminello, ACD co-founder and director of advocacy. "The addition of GAMT to the United States RUSP is a very important step that we believe will pave the way for GAMT to be included on all newborn screening panels worldwide, our ultimate goal."

About ACD: The Association for Creatine Deficiencies was established in 2012 with the mission to eliminate the challenges of living with cerebral creatine deficiency syndrome. ACD is committed to providing patient, family, and public education to advocate for early intervention through newborn screening, and to support and drive medical research for treatments and cures for CCDS. 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 creatineinfo.org.