ACD Announces the Advancement of Universal GAMT Newborn Screening for Approval by U.S. Secretary of Health

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Association for Creatine Deficiencies

Carlsbad, CA (May 12, 2022)--The Association for Creatine Deficiencies announced that after six years of advocacy efforts, the United States Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) voted unanimously today to advise the Secretary of Health and Human Services Xavier Becerra to add Guanidinoacetate Methyltransferase (GAMT) Deficiency 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.

> "This will expand awareness for all creatine deficiency syndromes that we hope one day will all be amenable to newborn screening and innovative therapies."
> *Dr. Nicola Longo*

GAMT was first nominated for review by the ACHDNC committee in spring 2016. After careful consideration, the committee decided that having an infant identified through a live population implemented screening would be necessary to prove the screening method's efficacy. Three infants have since been identified at birth through newborn screening testing.

"That initial vote against the recommendation to add GAMT to the RUSP was devastating. I can't help but wonder how many children's diagnoses have been missed in these six years, but I'm

looking to the future with hope for tomorrow's GAMT babies. Thank you to the Utah and New York programs that identified the first two babies and screened for GAMT when no one else would," commented ACD Co-Founder and Director of Advocacy Kim Tuminello.

ACD, along with Dr. Marzia Pasquali and Dr. Nicola Longo, re-nominated GAMT in spring 2021. In August 2021 the committee concluded that GAMT was deserving of a full evidence review which was to take place for the next nine months. ACD Scientific Medical Advisory Board members Dr. Pasquali, Dr. Longo, Dr. Andreas Schulze, and Dr. Saadet Andrews, as well as ACD Executive Director Heidi Wallis, were members of the GAMT Technical Expert Panel that provided consultation and advice to the evidence review committee throughout the process. "It was very encouraging to see a sincere interest in capturing the families' experiences in the evidence collected," Wallis said. "The review committee asked important questions and deferred to me, as a GAMT mom, to answer many of their concerns. The process was conducted in a fair and caring manner and I'm grateful to all parties involved and of course thrilled with the committee's decision."

"We are delighted to hear of the decision by the ACHDNC committee that, after a rigorous review process, has recommended the inclusion of GAMT deficiency in the uniform newborn screening panel" says Dr. Pasquali, an expert of newborn screening and biochemical genetic testing. Dr. Longo, a physician who diagnoses and treats many children with GAMT deficiency, added, "This will expand awareness for all creatine deficiency syndromes that we hope one day will all be amenable to newborn screening and innovative therapies."

Upon the Secretary of Health's approval of adding GAMT to the RUSP, each state in the U.S. and province, territory, state, and country around the world will decide if they will follow this advice to screen infants for GAMT. An addition of a disorder to the RUSP does not guarantee screening in most screening programs, but it raises the likelihood greatly. Several states in the U.S. have adopted legislation that commits their state to adding new RUSP disorders in a timely manner. Currently GAMT is screened in Utah, New York, British Columbia, Canada, and Victoria, Australia.

"I wish to give my deepest, heartfelt thanks to all the families whose children are affected by GAMT and have, despite the devastating challenges they face, risen up to share their story, advocate, and change the course of future generations' lives," stated Wallis.

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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