U.S. Secretary of Health Recommends Universal Newborn Screening for GAMT Deficiency

CARLSBAD, CA, UNITED STATES, January 16, 2023 /EINPresswire.com/ -- The Association for Creatine Deficiencies (ACD) announced that the United States Secretary of Health and Human Services Xavier Becerra has officially approved the addition of Guanidinoacetate Methyltransferase (GAMT) Deficiency to the Recommended Uniform Screening Panel (RUSP), recommending that all babies born in the United States be tested for GAMT as newborns. The RUSP provides state newborn screening programs with a carefully curated list of disorders that meet the committee's criteria for inclusion.

With the addition of 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 all 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 for in Utah, New York, and Michigan, Victoria, Australia, and British Columbia and Ontario, Canada.

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 such as autism and hyperactivity. 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.

ACD spearheaded advocacy efforts for the inclusion of GAMT on the RUSP for six years. GAMT was first nominated for inclusion on the RUSP in 2016 but was not approved due to the lack of one infant identified through a newborn screening pilot. In December 2020, that infant was identified by the Utah Public Health Laboratory's newborn screening program. GAMT was once again nominated in May 2021 and approved for evidence review in August 2021. The evidence review committee presented their findings May 12, 2022 and a unanimous vote by the Advisory Committee for Heritable Disorders in Newborns and Children (ACHDNC) recommended the addition of GAMT to the RUSP.

Heidi Wallis, ACD executive director and mother of two GAMT children, commented, "Many champions were involved in these efforts. We are grateful to each of the experts that gave of

their time and expertise in the nomination and evidence review process, the parents that testified at ACHDNC and state meetings, and ultimately the ACHDNC members and Secretary Becerra for their decision to add GAMT to the RUSP. Most importantly, we wish to acknowledge the individuals affected by GAMT and their families. We hope this milestone is marked with a feeling of relief, hope, and justice in the world.

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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