ASSOCIATION FOR CREATINE DEFICIENCIES

BUILDING STRENGTH
changing lives

September 1, 2022

The Honorable Xavier Becerra
Secretary of Health and Human Services
U.S. Department of Health and Human Services
200 Independence Avenue, SW
Washington, D.C. 20201

Dear Secretary Becerra,

On May 12, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) unanimously voted and recommended that Guanidinoacetate Methyltransferase Deficiencies (GAMT) be added to the Recommended Uniform Screening Panel (RUSP). We are writing to ask that you approve this recommendation as quickly as possible, as this is a devastating and neurodegenerative disease.

GAMT is an inherited disorder that primarily affects the brain and muscles. Without early treatment, people with this disorder have neurological problems that are severe. These problems include intellectual disability, speech development limited to a few words, and recurrent seizures (epilepsy). Affected individuals may also exhibit autistic behaviors that affect communication and social interaction and self-injurious behaviors such as head-banging. Other features of this disorder can include involuntary movements (extrapyramidal dysfunction) such as tremors and facial tics. People with GAMT have weak muscle tone and delayed development of motor skills such as sitting or walking, and may lose previously acquired skills such as the ability to support their head or to sit unsupported.

GAMT is treated with incredibly safe, affordable, and effective supplements including creatine monohydrate and l-ornithine. Early treatment of younger siblings at birth has proven to allow these children to experience typical development. Newborn screening of these babies gives them the gift of a life free from disabilities. It also saves the families and caregivers of these children from financial and emotional hardships, and the government from financial responsibility.

The ACHDNC has done an extensive evidence review of GAMT, and are in agreement that this is a devastating disease that warrants the federal recommendation of newborn screening. The RUSP has proven to save lives every day, and we would like to respectfully request that you approve this as quickly as possible, so that no other child has to live with the unnecessary consequences of not having received immediate treatment.

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The Association for Creatine Deficiencies (ACD) board of directors, along with our scientific medical advisory board, are here to support those that are diagnosed with GAMT. If there are any further questions that we can help answer, please don't hesitate to contact us at kim@creatineinfo.org (801)893-0543.

We are excited for the future of GAMT children in the U.S., as we know that with your final approval, lives will be forever changed!

Sincerely,

Kim Tuminello
Co-Founder
Director of Advocacy
Genetics

Heidi Wallis
Executive Director

Dr. Nicola Longo MD, PhD
Chair, ACD Medical Advisory
Board Chair and Chief, Medical Pediatric
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