MDHHS adds Guanidinoacetate Methyltransferase Deficiency to newborn screening panel

LANSING, MICHIGAN, USA, September 19, 2022 /EINPresswire.com/ -- September is Newborn Screening Awareness Month, and starting this month Michigan babies with guanidinoacetate methyltransferase (GAMT) deficiency will now be diagnosed early thanks to the addition of a new screening to the state's newborn screening panel. GAMT is an inherited disorder that primarily affects the brain and muscles.

Newborn screening is a public health program required by Michigan law to identify babies with rare but serious disorders, like GAMT deficiency. All babies need to be screened in order to find the small number who look healthy but have a rare medical condition. Michigan's newborn screening system provides testing and follow-up for more than 50 conditions. Michigan is the third state in the United States to screen for GAMT deficiency.

"By being one of the first states to implement GAMT deficiency screening, Michigan continues to display its dedication to providing newborns the opportunity to achieve the best possible health outcomes," said Dr. Natasha Bagdasarian, MDHHS chief medical executive.

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.

Heidi Wallis, executive director of the Association for Creatine Deficiencies (ACD), personally knows the importance of early diagnosis and treatment. 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.

"I have two children with GAMT, and the difference a diagnosis at birth makes is nothing short of life-changing," Wallis said. "My daughter, who was diagnosed at 5-years-old, has recurrent seizures that cause her to sustain serious injuries. She is intellectually disabled and will need constant care the rest of her life. My son, diagnosed and treated at birth, is a neurotypical 10-

year-old and will no doubt become an independent and contributing member of society."

In May, ACD announced that after six years of advocacy efforts, the United States Advisory Committee on Heritable Disorders in Newborns and Children voted unanimously to advise the Secretary of Health and Human Services Xavier Becerra to add GAMT 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. GAMT is expected to be added to the RUSP upon HHS Secretary Becerra's approval in November.

"We are grateful for Michigan's addition of GAMT deficiency to their state newborn screening panel, and the momentum this will bring towards universal screening for GAMT," said Wallis.

To learn more about GAMT deficiency, visit babysfirsttest.org and creatineinfo.org.

To learn more about newborn screening in Michigan, contact the MDHHS Newborn Screening Program at 866-673-9939, via email at newbornscreening@michigan.gov or visit Michigan.gov/newbornscreening.

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