Guanidinoacetate methyltransferase (GAMT) Deficiency Overview
Newborn Screening Technical Advisory Committee
September 8, 2023

ABOUT THE CONDITION

- GAMT Deficiency is a rare inherited amino acid disorder that prevents the body from properly producing creatine, which helps your organs store and use energy.[1,2]
- Without enough creatine, the body's organs do not get enough energy to support its vital functions, which can cause damage.
- This damage primarily affects the brain and muscles, as these organs need the most energy.
- Without early treatment, GAMT deficiency can cause serious cognitive and neurological impairments.
- GAMT deficiency is caused by changes in the GAMT gene.

SIGNS & SYMPTOMS

- People with GAMT Deficiency may begin showing symptoms from early infancy to age three.
- Signs and symptoms can vary but may include mild to severe intellectual and developmental disabilities, delayed sitting or walking, delayed or limited speech ability, muscle weakness or low muscle tone, behavioral issues (anxiety, aggression, self-injury, hyperactivity), seizures, and uncontrollable movements.

DIAGNOSIS

- GAMT Deficiency can be detected through a newborn screening blood spot using tandem mass spectrometry.
- Diagnostic tests include testing for guanidinoacetate (GUAC) and low creatine in the blood after a positive newborn screening test.
- Molecular testing can also be helpful.

TREATMENT

- May include creatine and ornithine supplements, sodium benzoate (a medication that can reduce levels of an amino acid called glycine), medications to treat seizures, a lifelong diet low in protein, and speech, occupational, and behavior therapy.

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1. National Institutes of Health, Genetic and Rare Diseases Information Center. Guanidinoacetate methyltransferase deficiency - About the Disease. Updated February 2023. Accessed August 25, 2023. https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency

2. Health Resources & Services Administration. Guanidinoacetate methyltransferase deficiency | Newborn Screening. Updated June 2023. Accessed August 25, 2023. https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency