WASHINGTON STATE BOARD OF HEALTH

Washington State Department of HEALTH

Arginase 1 Deficiency (ARG1-D) Overview

ABOUT THE CONDITION

- ARG1-D is a rare and inherited metabolic disease that prevents the body from properly breaking down the amino acid arginine, an enzyme in the blood.[1,2]
- Arginase is one of six enzymes responsible for breaking down arginine and is part of an essential process in the body called the urea cycle.
- The urea cycle helps remove ammonia (or nitrogen) from the body, a waste product used to process protein.
- If the arginase enzyme isn't working properly, the body can't break down arginine and get rid of ammonia through the urea cycle.
- Irregularities in the urea cycle may cause levels of ammonia in the blood to increase.
- When ammonia levels become too high, it has toxic effects and can cause serious damage to the nervous system and other parts of the body.

SIGNS & SYMPTOMS

- Signs of ARG1-D can vary widely and may appear anytime from infancy to early childhood.
- Symptoms of ARG1-D include seizures, muscle tightness or stiffness, difficulty eating, vomiting, and trouble breathing.
- People with ARG1-D might also experience delays in both physical and cognitive development, loss of developmental milestones, and intellectual disabilities.

DIAGNOSIS

- ARG1-D can be detected through a newborn screening blood spot using tandem mass spectrometry.
- Diagnostic tests include testing for ammonia levels, amino acids, and urine organic acids (specifically orotic acid) after a positive newborn screening test.

TREATMENT

- May include a diet low in protein, special foods or formulas, eating regularly and avoiding missing meals, and medications to get rid of extra arginine and ammonia in the body.

To request this document in an alternate format or a different language, please contact the State Board of Health at (360) 236-4110 or by email at wsboh@sboh.wa.gov.

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1. Morales A, Sticco KL. Arginase Deficiency - NIH Bookshelf. In: *StatPearls*. StatPearls Publishing; 2023. Accessed August 25, 2023. http://www.ncbi.nlm.nih.gov/books/NBK482365/

2. Health Resources and Services Administration. Arginase deficiency | Newborn Screening. Updated June 2023. Accessed August 25, 2023. https://newbornscreening.hrsa.gov/conditions/arginase-deficiency

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Guanidinoacetate methyltransferase (GAMT) Deficiency Overview

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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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

Ornithine Transcarbamylase Deficiency (OTCD) Overview

ABOUT THE CONDITION

- OTCD is an inherited metabolic disorder that prevents the body from properly removing ammonia waste.[1,2]
- OTCD is an X-linked recessive condition, meaning it affects males more than females.
- OTC is an enzyme that is part of an essential process in the body called the urea cycle. The urea cycle removes ammonia (or nitrogen) from the body.
- People with OTCD do not make enough of the OTC enzyme.
- If the OTC enzyme isn't working properly, the body can't break down proteins during the urea cycle.
- Irregularities in the urea cycle may cause levels of ammonia in the blood to increase.
- When ammonia levels become too high, it has toxic effects and can cause serious damage to the nervous system and other parts of the body.

SIGNS & SYMPTOMS

- Signs of OTCD can vary widely and may appear anytime from infancy through adulthood.
- Symptoms of OTCD include seizures, floppy arms and legs (hypotonia), lack of energy, difficulty eating, vomiting, and trouble breathing.
- People with OTCD might also experience neurological and behavioral symptoms, intellectual disabilities, coma, or even death.

DIAGNOSIS

- OTCD can be detected through a newborn screening blood spot using tandem mass spectrometry.
- Diagnostic tests include testing for ammonia levels, amino acids, and urine organic acids (specifically orotic acid) after a positive newborn screening test.

TREATMENT

- May include a diet low in protein, special foods or formulas, amino acid supplements, medications and dialysis to get rid of ammonia in the body, and liver transplant.

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1. Donovan K, Vagar S, Guzman, N. Ornithine transcarbamylase deficiency- NIH Bookshelf. In: *StatPearls*. StatPearls Publishing; 2024. Accessed July 17, 2024. https://www.ncbi.nlm.nih.gov/books/NBK537257/

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