Arginase 1 Deficiency (ARG1-D) Overview
Newborn Screening Technical Advisory Committee
September 8, 2023

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.

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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