Genetics Uncoded:

NxGen MDx

Facts about Classic Citrullinemia

What Your Test Results Mean

Carriers typically show no signs or symptoms of classic citrullinemia, also called type I citrullinemia; however, carriers are at an increased risk of having a child born with classic citrullinemia. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Classic Citrullinemia Explained

Classic citrullinemia is an inherited metabolic disorder caused by excess nitrogen build up in the body. Symptoms include lethargy (lack of energy), vomiting, seizures, and poor feeding shortly after birth. Variations in the *ASS1* gene reduce or eliminate the enzyme argininosuccinate synthase 1, which is a critical enzyme in the urea cycle. Without proper activity from this enzyme, the body cannot process nitrogen properly. The excess nitrogen typically takes the form of ammonia and accumulates in the bloodstream. Ammonia is highly toxic to the body. A less common and milder form of the disease will cause clinical symptoms later in life and typically take the form of headaches, ataxia (issues with balance and muscle coordination), partial loss of vision, and lethargy. There is no cure for this disease, but patients can manage the disease by closely monitoring their diet as well taking medications that can decrease blood ammonia level.

How the Genetics Work

Classic citrullinemia is an autosomal recessive disorder caused by variants in the *ASS1* gene. In general, individuals have two copies of the *ASS1* gene. Carriers of classic citrullinemia have a variant in one copy of the *ASS1* gene while individuals with the disorder have variants in both copies of *ASS1*, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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