Citrin Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of citrin deficiency. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of citrin deficiency for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for citrin deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

Citrin Deficiency Explained

What is Citrin Deficiency?

Citrin deficiency or type II citrullinemia chiefly affects the nervous system, causing confusion, restlessness, memory loss, abnormal behaviors (such as aggression, irritability, and hyperactivity), seizures, and coma. Affected individuals often have specific food preferences, preferring protein-rich and fatty foods and avoiding carbohydrate-rich foods. The signs and symptoms of this disorder typically appear during adulthood and can be triggered by certain medications, infections, surgery, and alcohol intake. These signs and symptoms can be life-threatening in people with adult-onset type II citrullinemia.

Adult-onset type II citrullinemia may also develop in people who as infants had a liver disorder called neonatal intrahepatic cholestasis caused by citrin deficiency (NICCD). This liver condition is also known as neonatal-onset type II citrullinemia. NICCD blocks the flow of bile and prevents the body from processing certain nutrients properly. In many cases, the signs and symptoms of NICCD go away within a year. In rare cases, affected individuals develop other signs and symptoms in early childhood after seeming to recover from NICCD, including delayed growth, extreme tiredness, specific food preferences, and abnormal amounts of fats in the blood. This condition is known as failure to thrive and dyslipidemia caused by citrin deficiency (FTTDCD). Years or even decades later, some people with NICCD or FTTDCD develop the features of adult-onset type II citrullinemia.

Prognosis

Life expectancy depends on the severity of signs and symptoms for patients and their response to treatment. Symptoms may resolve on their own for affected infants. Some affected individuals have a poor prognosis due to liver cirrhosis. Liver transplantation may be considered for severe cases.

Treatment

Treatment is symptomatic as there is no cure for citrin deficiency. Liver transplantation can be considered to prevent hyperammonemic crisis, corrects metabolic disturbances, and eliminates preferences for protein-rich foods. Treatment focuses on reducing blood ammonia levels through diet, medication, and dialysis.

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Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/citrullinemia

National Urea Cycle Disorders Foundation
http://www.nucdf.org/

National Society of Genetic Counselor
https://www.nsgc.org/

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