Citrullinemia Type 1

What Your Results Mean

Test results indicate that you are a carrier of citrullinemia type 1. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of citrullinemia type 1 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 citrullinemia type 1, each of your children has a 1 in 4 (25%) chance to have the condition.

Citrullinemia Type 1 Explained

What is Citrullinemia Type 1?

Citrullinemia type 1 is an inherited metabolic disorder caused by excess nitrogen build up in the body. The excess nitrogen typically takes the form of ammonia and accumulates to toxic levels in the bloodstream. Infants appear normal at birth and start to show symptoms by the first week of life. Symptoms include lethargy (lack of energy), vomiting, seizures, pressure around the brain, and poor feeding. If left untreated, signs and symptoms can worsen and become life-threatening. A less-common, milder form of citrullinemia type 1 presents with symptoms later in life and causes headaches, ataxia (issues with balance and muscle coordination), partial loss of vision, and lethargy.

Prognosis

Prognosis is not well defined. If left untreated, citrullinemia type 1 can be fatal early in infancy. With treatment, individuals with citrullinemia type 1 can survive for an unknown amount of time, but treatment cannot reverse neurological impairment.

Treatment

Treatment is symptomatic as there is no cure for citrullinemia type 1. Treatment focuses on reducing blood ammonia levels through diet, medication, and dialysis.

Resources

National Urea Cycle Disorders Foundation
http://www.nucdf.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/citrullinemia-type-1
National Society of Genetic Counselors
https://www.nsgc.org/

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