Cystinosis

NxGen MDx

What Your Results Mean

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

Cystinosis Explained

What is Cystinosis?

Cystinosis is an inherited condition that prevents the body from breaking down the amino acid cysteine. Beginning in infancy, the accumulated cysteine forms crystals that damage the body's organs, especially the kidneys and eyes. Nephropathic cystinosis is the most severe type, and symptoms can begin during infancy. Symptoms of nephropathic cystinosis include kidney failure, increased urination and dehydration, soft bones, and acidic blood. If untreated, individuals experience kidney failure by the first decade of life. Intermediate cystinosis has the same signs and symptoms as nephropathic cystinosis; however, they begin later in life around adolescence. Affected individuals experience malfunctioning kidneys and corneal crystals. If intermediate cystinosis is left untreated, kidney failure occurs in the late teens or mid-twenties. Individuals with non-nephropathic or ocular cystinosis experience photophobia from corneal crystals but typically do not develop kidney malfunction. Age of onset for this type of cystinosis varies.

Prognosis

Untreated cystinosis severely reduces life expectancy, but proper medical treatment can improve an individual's survival into their fifties. Some people with cystinosis do not develop symptoms until adolescence, while others develop only photophobia with no kidney involvement.

Treatment

Cystinosis is a treatable disease. A kidney transplant is often required. A medication called cysteamine reduces the crystallization of cystine in the body's tissues.

Resources

Cystinosis Foundation
http://www.cystinosisfoundation.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/cystinosis/
National Society of Genetic Counselors
https://www.nsgc.org/

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