Genetics Uncoded:

Facts about Cystinosis

NxGen MDx

What Your Test Results Mean

Carriers show no symptoms of cystinosis and are not at risk to develop symptoms of the disorder. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Cystinosis Explained

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 cause damage to the body's organs, especially the kidneys and eyes. When the kidneys are damaged in this way, they eliminate important nutrients from the body, causing the affected child to have poor growth and soft bones. Cystinosis can eventually cause the kidneys to fail and cystine crystal formation in the eyes makes them extremely sensitive to light (photophobia). Cystinosis is a treatable disease; a medication called cysteamine reduces the crystallization of cystine in the body's tissues. Kidney transplant is often required. Untreated cystinosis severely reduces life expectancy for affected individuals, but proper medical treatment can allow them to live into their fifties. Some people with cystinosis do not develop symptoms until adolescence, while others develop only photophobia with no kidney involvement.

How the Genetics Work

Cystinosis is an autosomal recessive disorder caused by pathogenic variants in the *CTNS* gene. In general, individuals have two copies of the *CTNS* gene. Carriers of cystinosis have a single variant in one copy of the *CTNS* gene while individuals with cystinosis have variants in both copies of their genes—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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