Genetics Uncoded:

NxGen MDx

Facts about Congenital Finnish Nephrosis

What Your Test Results Mean

Carriers show no symptoms of congenital Finnish nephrosis 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.

Congenital Finnish Nephrosis Explained

Congenital Finnish nephrosis is an inherited condition that impairs the ability of the kidneys to filter protein out of the urine. This disease is usually diagnosed shortly after birth before 3 months of age. Affected infants have difficulty getting nutrients and swell with excess fluid. One of the important proteins lost in the urine of these patients is albumin. Albumin is a protein that acts like a sponge, drawing extra fluid from the body into the bloodstream where it remains until removed by the kidneys. When albumin leaks into the urine, the blood loses its capacity to absorb extra fluid from the body, causing edema. They also have compromised immune systems and are prone to infections. Congenital Finnish nephrosis is often fatal in infancy or early childhood; however, kidney transplant is curative. Without a transplant, affected individuals die by 5 years of age, but a successful transplant can allow for a normal lifespan.

How the Genetics Work

Congenital Finnish nephrosis is an autosomal recessive condition caused by pathogenic variants in the *NPHS1* gene. In general, individuals have two copies of the *NPHS1* gene. Carriers of congenital Finnish nephrosis have a single pathogenic variant in one copy of the *NPHS1* gene while individuals with congenital Finnish nephrosis have pathogenic 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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