What Your Test Results Mean

Testing results indicate that you are a carrier of steroid-resistant nephrotic syndrome. Carriers typically show no symptoms of steroid-resistant nephrotic syndrome; however, carriers are at an increased risk of having a child with steroid-resistant nephrotic syndrome. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

Steroid-Resistant Nephrotic Syndrome Explained

Steroid-resistant nephrotic syndrome is an inherited condition that impairs ability of the kidneys to filter protein out of the urine. This disease is usually diagnosed shortly after birth, and always 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. This disease leads to kidney failure that doesn’t respond to steroidal medications, and affected individuals usually require a kidney transplant before the age of 20. With careful medical care and a timely transplant, affected children can live well into adulthood.

How the Genetics Work

Steroid-resistant nephrotic syndrome is an autosomal recessive disorder caused by mutations in the NPHS2 gene. In general, individuals have two copies of the NPHS2 gene. Carriers of steroid-resistant nephrotic syndrome have a single mutation in one copy of the NPHS2 gene while individuals with steroid-resistant nephrotic syndrome have mutations in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Recommended Next Steps

Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?

Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.