Facts about Steroid-Resistant Nephrotic Syndrome

Steroid-Resistant Nephrotic Syndrome Explained

Steroid-resistant nephrotic syndrome is an inherited condition that impairs the ability of the kidneys to filter protein out of the urine. While a common disease risk factor, R229Q, is identified in 3-4% of the general population, it is typically only of concern if identified in combination with specific pathogenic \textit{NPHS2} variants. The severe form of the disorder 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. The disorder 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 variants in the \textit{NPHS2} gene. In general, individuals have two copies of the \textit{NPHS2} gene. Carriers of steroid-resistant nephrotic syndrome have a single variant in one copy of the \textit{NPHS2} gene while individuals with steroid-resistant nephrotic syndrome 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.