Nephrotic Syndrome/ Steroid-Resistant Nephrotic Syndrome, *NPHS2*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of nephrotic syndrome/steroid-resistant nephrotic syndrome. Carriers typically show no symptoms. Risk for 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 for 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 and their own personal clinical management.

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 nephrotic syndrome, each of your children has a 1 in 4 (25%) chance to have the condition.

Nephrotic Syndrome/Congenital Finnish Nephrosis Explained

What is Nephrotic Syndrome/Congenital Finnish Nephrosis?

Nephrotic syndrome, also known as steroid-resistant nephrotic syndrome, is an inherited condition that impairs ability of the kidneys to filter protein out of the urine. The age at which symptoms begin varies; in some cases, symptoms have begun before age two, while in others symptoms did not appear until later in childhood.

Symptoms include an excess of protein in the urine, a shortage of protein in the blood, an excess of cholesterol and triglycerides in the blood, and generalized swelling in the body tissues. The water-retention that causes swelling can also cause weight gain and high blood pressure. The disease can cause scar tissue to form in the kidneys' glomeruli, which are structures responsible for filtering waste products. This is known as focal segmental glomerulosclerosis.

The disease typically leads to kidney failure, necessitating transplantation in many before the age of 20. Even after receiving a kidney transplant, symptoms of the disease can recur. It is described as "steroid-resistant" because unlike other forms of nephritic syndrome, it does not respond to steroid medications.

The disease is caused by a variant in the gene that provides the instructions for making podocin, a protein used by the kidneys' glomeruli.

Prognosis

Prognosis for this condition is generally poor, as it is resistant to steroid treatment. The disease typically leads to kidney failure in the first or second decade, necessitating transplantation. Some individuals may live into adulthood after transplant, though disease recurrence can occur after transplant.

Treatment

Treatment for nephrotic syndrome typically requires kidney transplantation in the first or second decade, as the disease is not responsive to steroid therapy. Success of transplantation is variable.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022

Resources

National Kidney Foundation
https://www.kidney.org/

The NephCure Foundation
https://nephcure.org/

National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 4/2022