Nephrotic Syndrome/ Congenital Finnish Nephrosis, *NPHS1*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of nephrotic syndrome/congenital Finnish nephrosis. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of nephrotic syndrome/congenital Finnish nephrosis for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a 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.

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 are carriers for nephrotic syndrome/congenital Finnish nephrosis, 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/congenital Finnish nephrosis is an inherited condition that prevents the kidneys from filtering protein out of the urine. Symptoms of the disease begin in the first days or weeks after birth. Infants are often born prematurely with a low birth weight. High levels of protein in the blood, combined with kidney failure, cause the body to swell with excess fluid. Frequent infections and potentially harmful blood clots can also develop.

Prognosis

Prognosis is generally unfavorable without treatment. Intensive medical therapy combined with kidney transplantation greatly improves outcomes and survival. If left untreated, death usually occurs before the age of four years.

Treatment

Treatment involves intensive medical therapy to control bacterial infections combined with kidney transplantation. Successful kidney transplantation offers a good opportunity for survival with an acceptable quality of life for affected individuals.

Resources
National Kidney Foundation
https://www.kidney.org/
National Society of Genetic Counselors
https://www.nsgc.org/

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