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What is Nephrotic Syndrome, *NPHS2*- Related?
Nephrotic syndrome, *NPHS2*-related is an inherited disease typically characterized by progressive kidney disease in childhood which is resistant to treatment with steroids.[1] It is caused by a defect in the production of a protein called podocin that is essential for normal kidney function. Symptoms associated with nephrotic syndrome, *NPHS2*-relatedare attributed to the kidney's inability to maintain normal blood protein levels.[2]

What are the Symptoms of Nephrotic Syndrome, *NPHS2*- Related and What Treatment is Available?
Nephrotic syndrome, *NPHS2*-related typically has onset during childhood however symptoms may be present at birth or develop within the first year of life. Although rare, onset can occur in adulthood. Nephrotic syndrome, *NPHS2*-related is usually a progressive disorder that is resistant to steroid treatment and results in end-stage renal disease within the first or second decade of life. Atypical cases with milder symptoms and/or partial responsiveness to steroids have been reported. Signs and symptoms of nephrotic syndrome type 2 may include[1]:

- Proteinuria (high protein levels in urine)
- Hypoalbuminemia (low protein levels in the blood)
- Hyperlipidemia (high fat levels in the blood)
- Edema (swelling due to excessive fluid in body cavities)
- Resistance to steroid therapy
- Progression to end-stage kidney disease

Kidney transplant is typically curative; however, recurrence of kidney disease following transplant has been reported [3]

How is Nephrotic Syndrome, *NPHS2*- Related Inherited?
Nephrotic syndrome, *NPHS2*-related is an autosomal-recessive disease caused by mutations in the *NPHS2* gene.[3] An individual who inherits one copy of an *NPHS2* gene mutation is a "carrier" and is not expected to have related health problems. An individual who inherits two mutations in this gene, one from each parent, is expected to be affected with nephrotic syndrome type 2.

If both members of a couple are carriers, the risk for an affected child is 25% in each pregnancy. Therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at Risk for Nephrotic Syndrome, *NPHS2*-related?
Nephrotic syndrome type 2 can occur in any race or ethnicity; however, its incidence and carrier frequency are unknown.[4]

What does a positive test result mean?
If a gene mutation is identified, an individual should speak to a physician or genetic health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

What does a negative test result mean?
A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

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Where can I get more information?

- The Nephcure Foundation: http://www.nephcure.org/

References

1. Genetics Home Reference. *NPHS2*. Available at: http://ghr.nlm.nih.gov/gene/NPHS2. Accessed March 12, 2012.
2. Caridi G, Perfumo F, Ghiggeri GM. NPHS2 (Podocin) mutations in nephrotic syndrome. Clinical spectrum and fine mechanisms. *Pediatr Res*. 2005 May; 57(5 Pt 2): 54R-61R.
3. Caridi G, Trivelli A, Sanna-Cherchi S, Perfumo F, Ghiggeri GM. Familial forms of nephrotic syndrome. *Pediatr Nephrol.* 2010 Feb; 25(2):241-252.
4. Franceschini N, North KE, Kopp JB, McKenzie L, Winkler C. NPHS2 gene, nephrotic syndrome and focal segmental glomerulosclerosis: A HuGE review. *Genet Med*. 2006 Feb; 8(2): 63-75.

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