NxGen MDx

Alport Syndrome, *COL4A4*-Related

What Your Results Mean

Test results indicate that you are a carrier of Alport syndrome, *COL4A4*-related. Carriers are not expected to show symptoms of Alport syndrome, *COL4A4*-related. Some carriers of Alport syndrome are unaffected, while others develop a less severe condition called thin basement membrane nephropathy, which is characterized by blood in the urine. You and your partner would both have to be carriers of Alport syndrome, *COL4A4*-related for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Alport syndrome can be caused by changes in a variety of genes. Carrier testing for genes related to Alport syndrome of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

We recommend that you share and discuss this information with all of your health care providers. 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 are carriers for Alport syndrome, *COL4A4*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Alport Syndrome, *COL4A4*- Related Explained

What is Alport Syndrome, *COL4A3*-Related?

Alport syndrome, *COL4A4*-related is an inherited disorder caused by abnormal type IV collagen production. It is characterized by kidney disease, sensorineural hearing loss, and sometimes eye abnormalities such as cataracts. Blood in the urine and high levels of protein in the urine are typically the first signs of the disorder, followed by hearing loss. Kidney failure may occur in the teenage years but may not develop until as late as 40-50 years of age.

Prognosis

The prognosis for Alport syndrome, *COL4A4*-related depends on the severity of symptoms of the individual, which may vary. Affected individuals typically develop end-stage kidney failure by their fourth or fifth decade in life.

Treatment

Treatment for Alport syndrome, *COL4A4*-related is symptomatic. Individuals are typically given medication--such as an ACE inhibitor--to help control high blood pressure. Kidney dialysis or transplantation is available for those with end-stage renal disease. Additionally, individuals may benefit from a hearing aid or surgery to remove cataracts.

Resources

Alport Syndrome Foundation
http://alportsyndrome.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/alport-syndrome/
National Society of Genetic Counselors
https://www.nsgc.org/

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