NxGen MDx

# Alport Syndrome, X-Linked

## What Your Results Mean

Test results indicate that you are a carrier of X-linked Alport syndrome. Female carriers typically show no symptoms of X-linked Alport syndrome; however, some female carriers may have blood present in their urine which is typically only detectable with laboratory testing. Additionally, some female carriers may also have varying levels of hearing loss that occurs later in life. Around 40% of female carriers will experience kidney failure in late adulthood, around 80 years old. Female carriers are at an increased risk to have a child with X-linked Alport syndrome. Risk for the current or future pregnancies depends on the sex of the baby.

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

Risk for your female child to be a carrier is 50%; the risk to have a male with X-linked Alport syndrome is also 50%. In the absence of clinical symptoms, reflexive testing for male partners of carriers of this syndrome is typically not indicated due to the X-linked inheritance pattern. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

# Alport Syndrome, X-Linked Explained

## What is X-Linked Alport Syndrome?

X-linked Alport syndrome 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 (hematuria) 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. Because this is an X-linked condition, males are more severely affected than females.

## Prognosis

Prognosis for X-linked Alport syndrome 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 X-linked Alport syndrome 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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Revised: 7/2020