



Genetics Uncoded:



# Facts about Choroideremia



## What Your Test Results Mean

**Carriers typically show no symptoms of choroideremia; however, carriers are at an increased risk of having a child with choroideremia.**

Because this disease is X-linked, risk for offspring is dependent on the sex of the baby.

## ● Choroideremia Explained

Choroideremia is an inherited condition that causes the retina of the eye to break down over time. This causes progressive vision loss that usually begins before the individual reaches 20 years of age. Night vision is the first affected, followed by peripheral vision. The field of vision gradually shrinks, affecting a person's central vision at about age 40 or 50 and eventually leading to blindness. Recent clinical trial results for gene therapy to cure choroideremia are showing promise for a cure.

## ● How the Genetics Work

Choroideremia is an X-linked disorder caused by pathogenic variants in the *CHM* gene. Males have one copy of the *CHM* gene while females have two copies of the *CHM* gene. Affected males have a variant in the *CHM* gene. While females typically have two X-chromosomes, and therefore two copies of the *CHM* gene, males typically only have one X-chromosome, and therefore one copy of the *CHM* gene. Female carriers have a single variant in one copy of the *CHM* gene, and therefore have a 50% chance of transmitting the variant onto each child and a 50% chance of not transmitting the variant onto each child. Males who inherit the variant will be affected, and females who inherit the variant will be carriers and rarely develop symptoms of the disorder.

## Questions?

Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.