**Genetics Uncoded:**

**FACTS ABOUT CHOROIDEREMIA**

_AKA | Progressive Tapetochoroidal Dystrophy, CHM, Choroideremia, TCD, Choroidal Sclerosis_

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**What Your Test Results Mean**

Testing results indicate that you are a carrier of choroideremia. 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 the current or future pregnancies is dependent on the sex of the baby. Consultation with a genetic counselor is recommended for more detailed risk assessment.

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**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 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.

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**How the Genetics Work**

Choroideremia is an X-linked disorder caused by mutations 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 mutation in the CHM gene. Females with a mutation in one copy of the CHM do not typically show symptoms and are unaffected carriers. Affected males will not pass the disorder onto their sons; however, daughters of affected males will be carriers of the disorder. Risk is 50% for sons of female carriers to inherit choroideremia. Risk is 50% for daughters of female carriers to also be female carriers. Rarely, females carriers will develop symptoms of choroideremia.

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**Recommended Next Steps**

Consultation with a genetic counselor for more detailed risk assessment is recommended.

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**Questions?**

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