

Choroideremia affects primarily males. Female carriers generally have few or no symptoms. However, a small number of females develop the disorder as a result of a genetic process that inactivates the normal gene and leaves only the dysfunctional gene active. In the Salla area of northern Finland, an unusually high number of people have been diagnosed with choroideremia; approximately one in forty persons. A doctor will perform tests that examine the patient's visual field suspected of having choroideremia and will look inside the eye for degeneration of the retina. Genetic testing is available for some genetic variants that cause choroideremia. The symptoms of choroideremia can be treated but the disease itself cannot yet be cured. Organizations providing services to sight impaired people help patients and their families. Genetic counseling is recommended for families affected by this disorder.