

What is choroideremia?

Choroideremia is a rare inherited disorder that causes progressive loss of vision due to degeneration of the choroid and retina.

What are the symptoms?

Choroideremia occurs almost exclusively in males. In childhood, night blindness is the most common first symptom. As the disease progresses, there is loss of peripheral vision or “tunnel vision” and later a loss of central vision. Progression of the disease continues throughout the individual’s life, although both the rate and the degree of visual loss can vary, even within the same family.

Vision loss due to choroideremia is caused by degeneration of several layers of cells that are essential to sight. These layers, which line the inside of the back of the eye, are called the choroid, the retinal pigment epithelium (RPE), and the photoreceptors. The choroid consists of several blood vessel layers located between the retina and the sclera (the “white of the eye”). Choroidal vessels provide the RPE and photoreceptors with oxygen and nutrients necessary for normal function. The RPE provides essential support functions for photoreceptors.

The photoreceptors are responsible for converting light into the electrical impulses that are transferred to the brain where images are created and “seeing” actually occurs.

Is it an inherited disease?

Choroideremia is genetically passed through families by the X-linked pattern of inheritance. In this type of inheritance, the gene for the disease is located on the X chromosome. Females have two X chromosomes and can carry the disease gene on one of their X chromosomes. Because they have a healthy version of the gene on their other X chromosome, carrier females are usually not affected by X-linked diseases. Males have only one X chromosome (paired with one Y chromosome) and are therefore genetically susceptible to X-linked diseases. Males with X-linked diseases pass their Y chromosome to their sons, and therefore will never pass an X-linked disease to their sons. Female carriers have a 50 percent chance (or 1 chance in 2) of passing the X-linked disease gene to their daughters, who become carriers, and a 50 percent chance of passing the gene to their sons, who are then affected by the disease.

CHOROIDEREMIA, Continued

Choroideremia is one of the few retinal degenerative diseases that might be detected prenatally in some cases; female carriers may want to seek information about this testing from a medical geneticist or a genetic counselor. All members of an affected family are encouraged to consult an ophthalmologist and to seek genetic counseling. These professionals can provide explanations of the disease and the recurrence risk for all family members and for future offspring. Genetic counselors are excellent resources for discussing inheritability, family planning, genetic testing, and other related issues.

What treatment is available?

Scientists have discovered that mutations in the choroideremia gene (CMH) gene cause choroideremia. In 2011, a research team in the United Kingdom (UK) launched a human study of a gene therapy for people affected by the disease. The treatment works by replacing bad copies of CHM with good copies. In 2014, the UK researchers reported that five of the first six participants in the trial had improved vision. In 2016, the investigators reported that those vision improvements had been sustained for 3.5 years. Researchers in the United States and Canada are also conducting choroideremia gene therapy clinical trials. Other emerging treatment approaches — including stem cells and pharmaceuticals — may also someday benefit people with choroideremia.

Until a treatment is discovered, help is available through low-vision aids, including optical, electronic, and computer-based devices. Personal, educational, and vocational counseling, as well as adaptive training skills, job placement, and income assistance, are available through community resources.

Are there any other related diseases?

Early in the course of the disease, choroideremia could be confused with X-linked retinitis pigmentosa. Both have symptoms of night blindness and tunnel vision. However, differences are clear with genetic testing and/or a comprehensive eye examination, especially as the disease progresses. The disease most similar clinically to choroideremia is gyrate atrophy, which is inherited recessively and caused by mutations in the ornithine aminotransferase gene.

Low-vision resources and extensive information on research and clinical trials for choroideremia are available at: www.FightingBlindness.org