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My46 Trait Profile

Choroideremia

Choroideremia is a rare genetic condition that causes night blindness in males at a young age, which progresses to tunnel vision. Central vision is typically maintained until late in life. It is caused by mutations in the REP-1 gene that makes a protein called Rab escort protein-1 (REP-1).

Characteristics of Choroideremia

Choroideremia is a rare eye disease that affects 1 in 50,000 to 100,000 males. It is characterized by the degeneration of the choroid (layers of cells that provide nourishment to the back of the eye), retinal pigment epithelium (RPE; located at the back of the eye) and photoreceptors (cells located in the retina that respond to light). Photoreceptors are light-sensitive cells and are responsible for vision. There are two types of photoreceptors: rods and cones. Rods are responsible for outer (peripheral) vision and the ability to see at night. Cones are responsible for central vision and color perception. Of the two types, rods are primarily affected by choroideremia.

In affected males, night blindness (difficulty seeing in dimly lit environments or nyctalopia) develops at a young age and progresses to tunnel vision (restriction of the field of vision). This means boys can appear clumsy, tripping or bumping into things that would typically be within the field of vision of someone without choroideremia. Central vision is typically maintained until age 40-50 years. The rate of progression in affected individuals can vary even within the same family. Rarely, the progression of vision loss can be worsened by invasion of new blood vessels growing from the choroid into the space under the RPE (choroidal neovascularization).

Carrier females generally have no visual impairment and normal visual fields, however signs of degeneration can be detected by a careful eye examination. Of note, there have been cases where carrier females develop milder symptoms of the condition compared to males, and a few cases where symptoms are as severe as males.

Diagnosis/Testing

The clinical diagnosis of choroideremia can be made based on an examination of the back of the eye (fundus) by a medical doctor who is specialized in eye and vision care (ophthalmologist) or an optometrist familiar with this condition and by family history evaluation. Visual testing by electroretinography (ERG) and/or optical coherence tomography (OCT) can also help determine the diagnosis. ERG testing evaluates retinal cell function and can reveal rod-cone degeneration whereas OCT imaging can reveal characteristic physical changes in the retinal and choroidal cell layers.

Genetic testing can confirm the clinical diagnosis by detecting a change or mutation in a gene called REP-1. This gene makes the Rab escort protein-1 which is involved in protein transport within the cells of the retina.

Management/Surveillance

Management of choroideremia involves regular eye exams by an ophthalmologist. These visits improve quality of life as much as possible, allowing for vision assessments, updated refractions (to determine the need for corrective lenses), and low vision evaluations as needed. A diet rich with fruits and green leafy vegetables and/or antioxidant vitamin supplementation as well as regular intake of dietary omega-3 very-long-chain fatty acids like docosahexaenoic acid (found in cold water, fatty fish like salmon) may be helpful. Smoking should be avoided.

Mode of inheritance

Choroideremia is inherited in an X-linked recessive pattern. This means the REP-1 gene is located on the X chromosome. Females have two X chromosomes, and males have one X chromosome and one Y chromosome. Males with a change or mutation in the REP-1 gene will be affected with choroideremia, while females with a mutation in the REP-1 gene on only one copy of their two X chromosomes are considered carriers of choroideremia and typically do not develop symptoms.

Risk to family members

The risk for a carrier mother to pass on her REP-1 gene mutation to her children is 50% with every pregnancy. If that child is male, he will be affected with choroideremia; while if that child is female, she will be a carrier.

Special considerations

Choroideremia can often be confused with another condition called retinitis pigmentosa, especially during the early course of the disease.

Resources

Choroideremia Research Foundation Canada
http://www.choroideremia.ca
Foundation Fighting Blindness Canada
http://www.ffb.ca
Genetics Home Reference: Choroideremia
http://ghr.nlm.nih.gov/condition/choroideremia

References

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