WHAT YOU SHOULD KNOW ABOUT CHOROIDEREMIA

WHAT IS CHOROIDEREMIA?

Choroideremia is an inherited retinal disease that causes progressive vision loss due to cell degeneration of the choroid, the retinal pigment epithelium (RPE), and the photoreceptors.

The retina is a thin piece of tissue lining the back of the eye, making vision possible.

The choroid consists of blood vessel layers located between the retina and the sclera.

The photoreceptors are responsible for converting light into the electrical impulses that are transferred to the brain.

The retinal pigment epithelium (RPE) provides essential support functions for photoreceptors.

WHAT TO EXPECT WITH CHOROIDEREMIA:

Night blindness is the most common first symptom

As the disease progresses, there is loss of peripheral vision

Later there is a loss of central vision as well

HOW IS CHOROIDEREMIA INHERITED?

Choroideremia is caused by mutations in the CHM gene. The condition is passed down in families by the X-linked pattern of inheritance.

Female carriers have a 50% chance of passing the disease gene to their daughters, who become carriers, and a 50% chance of passing the gene to their sons, who are then affected by the disease.

Males with X-linked diseases pass their Y chromosome to their sons, and therefore will never pass an X-linked disease to their sons. However, affected males will always pass their affected chromosomes to their daughters who will always become carriers.

As an X-linked disease, choroideremia occurs primarily in males.

Progression of the disease continues throughout the individual’s life.

Both the rate and the degree of visual loss can vary, even within the same family.

To learn more about living with choroideremia, finding a specialist, genetic testing or clinical trials, visit FightingBlindness.org or call 888-332-3667.