Treatment

There are no currently approved treatments or cure for CHM. However, clinical trials and other promising research in gene therapy, stem cell therapy, optogenetics and other targeted therapeutic treatments are underway. Individuals wishing to participate in these clinical trials or research studies will need to have their diagnosis of CHM verified with a genetic test.

Choroideremia
RESEARCH FOUNDATION

The Choroideremia Research Foundation (CRF) was formed in 2000 as a 501(c)3 public charity with the urgent mission to cure blindness caused by choroideremia (CHM). Since then, CRF has funded over $4 million in research grants and has become the largest nonprofit organization in the world exclusively dedicated to CHM.

CRF's mission includes patient education and advocacy and coordination of a global scientific coalition, the International Choroideremia Research Network. CRF hosts international conferences, scientific symposia, regional meetings; webinars; virtual and social support groups; and a robust website with patient, family and vision professional resources.

800-210-0233
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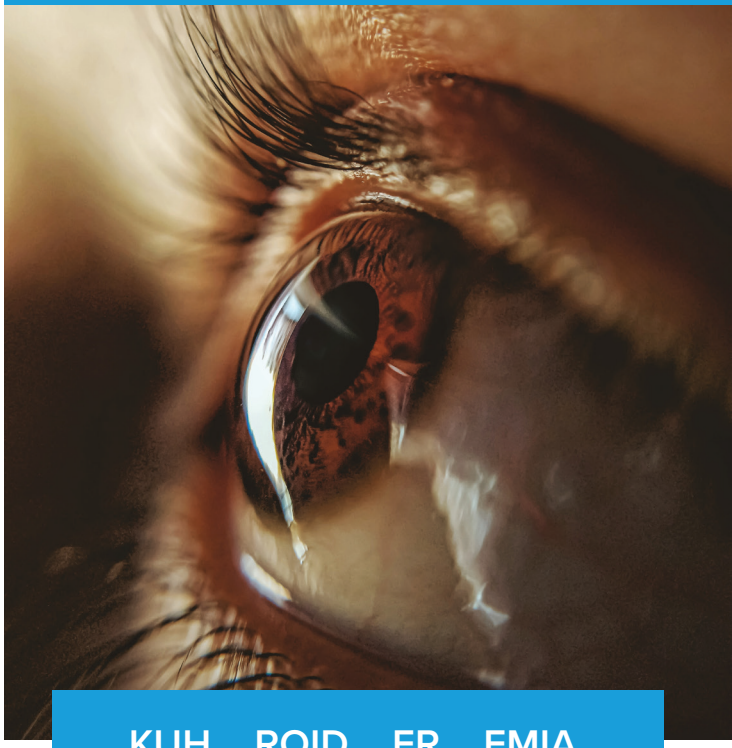
23 E. Brundreth St.
Springfield, MA 01109-2110 USA

Follow us @cureCHM on:

Learn more at www.curechm.org

What is Choroideremia?



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Choroideremia (CHM) is a rare inherited disorder that causes progressive vision loss, which may ultimately lead to complete blindness.

Choroideremia
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Cause

CHM is caused by a defect on the CHM gene, which lies on the X Chromosome. Due to this defect, the body does not produce a protein called RAB Escort Protein-1, or REP1. This protein is essential for the retina cells to function properly, without it, they eventually die.

CHM affects males fully at an early age, while female symptoms are less severe.

Symptoms

The first symptom of choroideremia (CHM) is generally night-blindness, followed by vision loss in the mid-periphery, or side edges. These "blind spots" appear in an irregular ring, leaving only patches of peripheral vision, while central vision is maintained. Late stage patients also experience more significant glare sensitivity, color and depth perception loss. Over time the peripheral vision loss extends in both directions leading to "tunnel vision" and progressive sight loss.

CHM is a rare disease affecting an estimated 1 in 50,000 individuals.

Inheritance

As CHM is carried on the X chromosome, a carrier female will have a 50/50 chance of a son being affected or a daughter being a carrier. For an affected CHM male, his daughter will be a carrier with 100% certainty; however, his son will not have CHM.

Diagnosis

A doctor will perform tests that examine the patient's visual field, light sensitivity, and look inside the eye for retinal degeneration. Various types of imaging equipment are used as a guide; however, CHM often presents with similar symptoms and exam images to another more common retinal degenerative condition called retinitis pigmentosa (RP). Both conditions cause similar damage, yet the root cause is different. A genetic test is the only way to know what is causing the vision loss. Testing may be available for free or covered by insurance. It is important to know the correct cause of vision loss as new treatment options in development target specific mutations, and what works to treat an RP patient, may not work for the CHM mutation and vice versa.

More information about genetic testing is available on CRF's website at **curechm.org.** If one member of a family tests positive for choroideremia, it is recommended that all other known family members who may have inherited the CHM gene also get tested.

What Else Can I Do?

Practice overall good eye care, wear 100% UVA & UVB protection wraparound sunglasses and brimmed hats.

Eat a nutritious diet rich in leafy green vegetables and foods with high antioxidant levels, exercise regularly, and take physician-approved nutritional supplements – all of these actions may help to delay CHM progression and protect retinal health.

Take advantage of all the resources the CRF has to offer by visiting **www.curechm.org:**

- Sign up to receive e-newsletters
- Locate a retinal specialist near you
- Follow/browse CRF's social media channels
- Connect with other family members
- Become a CRF member, participate as an event attendee, volunteer for a committee, fundraise or donate.

Taking these steps will help CHM patients and family members become more knowledgeable, confident, and empowered about their healthcare decisions.