

CHMstandards.org: Toward Developing Patient-Relevant Outcome Measures for Choroideremia Trials

"With a prevalence estimated at 1:50,000 approximately 6,000 men in the US experience progressive sight loss due to Choroideremia (CHM). CHM is a rare x-linked inherited disease caused by mutations in the REP1 gene and primarily affects males with the most debilitating manifestation being progressive loss of visual field. 100% of affected males will experience severe bilateral retinal degeneration and loss of visual field over their lifetimes. Women usually exhibit much milder symptoms but in some cases experience severe sight loss. Reduced night vision is usually the first noticeable symptom in early childhood with noticeable visual field loss in adolescence. The majority of visual field usually is lost by the third decade. There are currently no available treatment options for CHM patients.

This work reviewed available data and literature to inform development of optimal clinical trial designs and endpoints for therapies incorporating the patient perspective. Preservation of photoreceptors and supporting cells in the retina is an essential attribute for CHM therapies. This can be objectively measured with non-invasive imaging technology that is widely available. Therapeutic intervention to maintain sight can be beneficial to patients with any remaining vision. CHM is a rare pediatric disease and early intervention in children could prevent life-changing debilitation. We have developed a dynamic and collaborative online resource at [www.chmstandards.org](http://www.chmstandards.org).

This work was authored in collaboration with the Choroideremia Research Foundation by Randy Wheelock, BBA, Patient Advocate and Chief Advisor for Research and Therapy Development for the CRF, Ioannis (John) Dimopoulos, MD, MSc, Department of Ophthalmology, University of Ottawa, Paul Freund, BSc, MSc, MD, Department of Ophthalmology, University of Alberta, and Ian MacDonald, MD, CM, FCCMG, FRCSC, FCAHS, Department of Ophthalmology and Visual Sciences, University of Alberta.

The Choroideremia Research Foundation was founded in 2000 as a fundraising and patient advocacy organization to stimulate and support research on CHM. Since its inception the CRF has provided funding for basic research and preclinical development projects laying the groundwork for CHM gene therapy programs. The CRF's early and sustained investments have led to two currently active clinical trial programs and other preclinical programs expected to advance to human trials. The CRF also invests in research for other therapeutic approaches such as drug discovery and cell based therapies that may maintain, or possibly restore, sight. CHM was of the earliest ocular gene therapies to enter human trials and has provided a stepping stone for advances in gene therapy for other diseases. For more information please visit [www.curechm.org](http://www.curechm.org)."

Contact: Corresponding author: [randywheelock@curechm.org](mailto:randywheelock@curechm.org)