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CHOROIDEREMIA RESEARCH FOUNDATION EXPANDS SUPPORT FOR PROMISING RESEARCH PATH FOR RARE INHERITED RETINAL EYE DISEASE

Twin brothers have identical CHM mutation but one brother has no symptoms

SPRINGFIELD, MA, UNITED STATES, June 20, 2023 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce that Ian MacDonald, MD, CM in the Department of Ophthalmology and Visual Sciences at the University of Alberta is the first recipient of the Francisco Rodriguez Research Award. The research award is $20,558.

The award extends current research of Dr. MacDonald's, Identifying the cause of a discordant phenotype in 2 brothers with identical choroideremia (CHM) mutation. In the first research phase, he sought to explain why the same pathogenic mutation (c. 1359C>T) found in both brothers does not result in clinical symptoms in one brother who should have (significantly) developed them. In the next research phase, his team aims to identify the reason for the phenotype disparity through the establishment of patient-derived retinal pigment epithelial cell lines and development of more sensitive functional assays.

> Dr. MacDonald's research continues to explore promising paths to increase the research community's knowledge and understanding of how CHM functions."
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> *Neal Bench, CRF board president*

"Dr. MacDonald's research continues to explore promising paths to increase the research community's knowledge and understanding of how CHM functions," said Neal Bench, CRF board president.
For more information about all research studies the CRF supports, please visit curechm.org/research/

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About Choroideremia
Choroideremia (CHM) is a rare inherited form of blindness affecting approximately 1 in 50,000 people. Due to its x-linked inheritance pattern males are most severely affected with females usually experiencing much milder visual impairment. Symptoms begin in early childhood with night blindness and restriction of visual field being the earliest noticeable effects, eventually progressing to complete blindness. An estimated 6,000 people in the United States and 10,000 in the European Union are impacted by choroideremia. There are currently no approved treatments for choroideremia. For more information, visit curechm.org


Ian MacDonald, MD, CM

About the Choroideremia Research Foundation Inc.
The Choroideremia Research Foundation was founded in 2000 as an international fundraising and patient advocacy organization to stimulate research on CHM. Since its inception, the CRF has provided approximately $5 million in research awards and is the largest financial supporter of CHM research worldwide. Research funded by the CRF has led to the development of a CHM mouse model, the pre-clinical production of gene therapy vectors currently in clinical trials, and the CRF Biobank which stores tissue and stem cell samples donated by CHM patients. For more information, visit curechm.org

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