CHOROIDEREMIA RESEARCH FOUNDATION SUPPORTS NEW RESEARCH PATH FOR RARE INHERITED RETINAL EYE DISEASE

Dr. Ian MacDonald applies AON theory to stop aberrant splicing in CHM gene mutations

SPRINGFIELD, MA, UNITED STATES, May 23, 2022 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce its latest research award to Ian MacDonald, MD, CM in the Department of Ophthalmology and Visual Sciences at the University of Alberta. This award supports research into a new area of scientific study for choroideremia (CHM) that applies antisense oligonucleotide (AON) therapy.

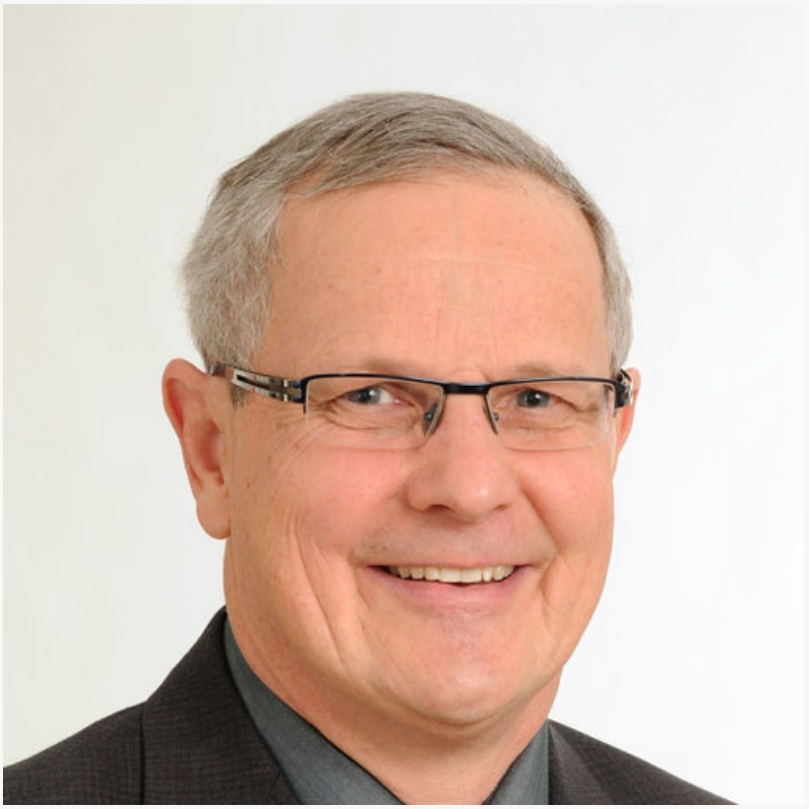
> Dr. MacDonald's research is a promising new path to treat CHM that would potentially provide an additional treatment option for patients."
>
> *Neal Bench, CRF board president*

In Dr. MacDonald's study titled, Developing an antisense oligonucleotide therapy for choroideremia, he hypothesizes that antisense oligonucleotide (AON) therapy may be applicable to a select number of CHM patients. CHM causes degeneration of the photoreceptors, retinal pigment epithelium (RPE) and choroid. All known mutations in the CHM gene result in absence of its protein product, Rab escort protein-1 (REP1). These mutations create new splice sites causing the inclusion of additional sequence in the mRNA and the production of an aberrant gene product. An AON therapy can suppress this aberrant splicing and restore a full-length normal transcript and functional protein. This approach offers a new direction for potential therapy with adeno-associated viral vectors.

Currently, Dr. MacDonald's center follows two unrelated patients who possess the same CHM causing intronic mutation. In preliminary experiments using patient-derived fibroblasts, four AONs were designed to induce skipping of the mutant exonic sequence. Of four trialed sequences, one showed an exceptional level of effectiveness in re-establishing near wild-type expression levels of normal CHM transcript and REP-1 protein. His team seeks to potentially develop a Precision Medicine therapy for these patients and explore antisense therapy as a treatment for CHM.

"Dr. MacDonald's research is a promising new path to treat CHM that would potentially provide

an additional treatment option," said Neal Bench, CRF board president. "We are proud to support his team's work by jointly funding it with the Choroideremia Research Foundation of Canada."


Ian MacDonald, MD, CM

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

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 over $2.5 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 animal 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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