CHOROIDEREMIA RESEARCH FOUNDATION EXPANDS RESEARCH SUPPORT INTO NONSENSE MUTATIONS OF A RARE INHERITED RETINAL DISEASE

Focus on ADAR-based RNA editing in the retina

Dror Sharon, PhD, Hadassah-Hebrew University Medical Center

SPRINGFIELD, MA, UNITED STATES, May 1, 2023 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce its latest scientific research grant to Dror Sharon, PhD with the Division of Ophthalmology, Hadassah-Hebrew University Medical Center (Jerusalem, Israel) and Shay Ben Aroya, PhD with Faculty of Life Sciences, Bar-Ilan University (Israel). The award amount is $74,152.

Mutations in choroideremia (CHM), encoding for Rab escort protein-1 (REP1), are known to cause CHM. Approximately 30% of CHM mutations are nonsense and as such, therapies that target nonsense mutations could be beneficial for a substantial proportion of CHM patients. In the investigators study, In-vitro and in-vivo RNA editing of CHM nonsense mutations, there are three aims to identify and study the efficacy of guide RNAs (gRNAs). They will target two relatively common CHM nonsense mutations, utilizing and recruiting endogenous cellular adenosine deaminase acting on RNA (ADAR) enzymes to perform editing and correction at the RNA level.

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> *Neal Bench, CRF board president*

- AIM 1 – use a yeast-based system as well as bioinformatic tools to identify the most efficient gRNAs for two CHM nonsense mutations (c.1218C>A, p.Cys406* and c.877C>T, p.Arg293*).
- AIM 2 – study the level of RNA editing of these mutations in HeLa cells overexpressing either ADAR1 or ADAR2 using a dual-reporter plasmid developed in the investigators’ lab as well as next generation sequencing of RT-PCR products.

• AIM 3 – harness endogenous photoreceptor and retinal pigment epithelium (RPE) ADAR enzymes, and especially ADAR2, to edit the studied CHM mutations in vivo.


Shay Ben Aroya, PhD, Bar-Ilan University

"Drs. Sharon and Aroya will enrich our knowledge on the efficacy of safety ADAR-based RNA editing, a first step towards RNA editing treatment in CHM patients," said Neal Bench, CRF board president.

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 about all research studies the CRF supports, please visit curechm.org/research/

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About Choroideremia
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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 nearly $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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