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

Dr. David Gamm uses Ace-tRNA readthrough therapy for CHM stop codon/nonsense mutations

Dr. David Gamm, University of Wisconsin, Madison

SPRINGFIELD, MA, UNITED STATES, April 27, 2022 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce the recipient of The Einhorn Family Research Award is David Gamm, MD, PhD, Director of the McPherson Eye Research Institute and Associate Professor of Ophthalmology and Visual Sciences at University of Wisconsin, Madison. This award supports research for which Dr. Gamm recently received an award from the Million Dollar Bike Ride 2021 Pilot Grant Program. Together, they amplify the impact of his research proposal with total funding of $124,360.

In Dr. Gamm's proposal, Ace-tRNA readthrough therapy for choroideremia caused by nonsense mutations, the aim is to optimize the delivery of anticodon engineered transfer RNA (ace-tRNA) in retinal pigmented epithelium (RPE) and photoreceptors (PRs) and test its efficacy. In choroideremia (CHM), mutations in the X-linked CHM gene are the underlying cause of the disease. Nonsense mutations leading to premature termination codons (PTCs), also known as stop codons, affect approximately 35% of CHM patients. The CHM gene encodes Rab Escort Protein-1 (REP-1) and its loss-of-function in the retina is responsible for disease progression. It is therefore presumed that restoring REP-1 activity in RPE and/or PRs prior to their complete degeneration will halt the disease process and preserve or restore retinal and visual function.

> “Dr. Gamm's proposal investigates other approaches for treating CHM, exploring new research paths that expand the scientific landscape.”
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> *Neal Bench, CRF board president*

The lack of appropriate disease models for CHM makes it challenging to study or develop therapeutic strategies. To overcome this issue, the Gamm lab has developed human induced pluripotent stem cell (hiPSC)-

derived RPE cell- and retinal organoid (RO)-based models for CHM. Dr. Gamm will use these models to test a novel tRNA-based readthrough technology as a potential therapeutic for CHM.

“Dr. Gamm is a recognized leader in the field of inherited retinal diseases who the CRF has supported in previous research efforts,” said Neal Bench, CRF board president. “His proposal investigates other approaches for treating CHM, exploring new research paths that expand the scientific landscape.”

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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