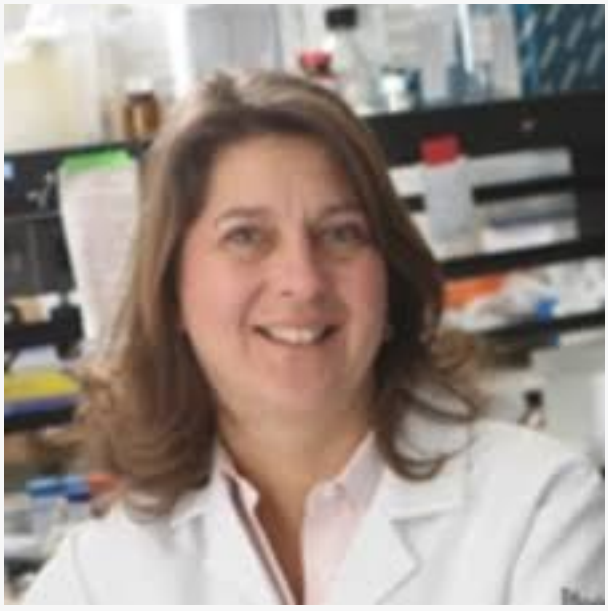
CHOROIDEREMIA RESEARCH FOUNDATION ACCELERATES SCIENTIFIC RESEARCH OF RARE INHERITED RETINAL DISEASE WITH THREE GRANTS

The foundation names it's 4th annual Randy Wheelock Award winner

SPRINGFIELD, MA, UNITED STATES, September 17, 2024 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce its latest scientific research grants, supporting its mission to find a treat or cure for choroideremia (CHM). Award recipients are as follows:

David Gamm, MD, PhD



Kathleen Boesze-Battaglia, PhD

Kathleen Boesze-Battaglia, PhD, Department of Basic and Translational Science, University of Pennsylvania

STUDY: Assessment of Metabolic Homeostasis in the CHMnull/wt mouse, implications for novel therapeutic interventions

SPECIFC AIMS:

To test the hypothesis that loss of REP1 expression compromises RPE metabolic homeostasis leading to RPE dysfunction resulting in nutrient deprivation to the neural retina and retinal degeneration.

1. To determine if loss of CHM/Rep-1 dysregulates systemic, retinal and RPE metabolism.
2. To determine if loss of CHM/Rep1 modulates RPE lipoprotein secretion and lipid transporter levels and activity.

GRANT: $59,310

David Gamm, MD, PhD (PI/PD), Director of McPherson Eye Research Institute, Professor in Department of Ophthalmology and Visual Sciences, University of Wisconsin-Madison

STUDY: Optimization of AAV vectors for ace-tRNA readthrough therapy for choroideremia

SPECIFIC AIMS:

To develop AAV vectors for efficient and long-term expression of ace-tRNAArg for readthrough of Arg>TGA mutations in CHM.

1. Generation of next-generation versions of CCT, TCG, and TCT 4X ace-tRNAArg constructs and corresponding scAAVs.
2. Assessment of the dose response of next-generation ace-tRNAArg scAAVs (from Aim 1) in CHM (Arg270X)-3XFLAG iPSC-RPE.
3. Determination of the long-term efficacy of optimized 4X ace-tRNAArg scAAV7m8.
4. Evaluation of the versatility of optimized 4X ace-tRNAArg scAAV using a second iPSC-RPE model of CHM harboring an Arg293X PTC.

GRANT: $100,000

Shalhevet Izraeli, MSc Student, Hadassah-Hebrew University Medical Center, Jerusalem, Israel

Ms. Izraeli is this year's Randy Wheelock Research Award Winner in celebration of the decades-long advocacy of its namesake to find treatment or a cure for CHM.

STUDY: Using suppressor transfer RNAs to correct CHM nonsense mutations

SPECIFIC AIMS:

To identify and study the translational readthrough efficacy of different suppressor transfer RNA (Sup-tRNA) targeting two relatively common CHM nonsense mutations to produce full length proteins with WT amino acids.

1. Use the HeLa and the 661W mouse photoreceptor cell-lines to identify the most efficient Sup-tRNA for two CHM nonsense mutations (c.877C>T, p.Arg293* and c.1218C>A, p.Cys406*), using a dual-reporter plasmid developed in our lab.
2. Clone multiple copies of the most efficient Sup-tRNA (identified in AIM1) for each mutation to find the desired copy number of Sup-tRNA required to produce sufficient levels of the full-length protein, using fluorescent microscopy, Western blot and FACS.

GRANT: $50,000

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/#choroideremia

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 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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