CHOROIDEREMIA RESEARCH FOUNDATION SUPPORTS RESEARCH INTO CRITICAL PROTEIN RELATED TO RARE INHERITED RETINAL EYE DISEASE

Dr. Vasiliki Kalatzis focuses on the dysregulation of ion channels linked to RPE

SPRINGFIELD, MA, UNITED STATES, June 6, 2022 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce its latest research award to Vasiliki Kalatzis, PhD who serves as Research Director at Inserm, Institute for Neuroscience, Montpellier, France. Her research will expand the scientific community's understanding about the absence of Rab escort protein-1 (REP1) and its connection to choroideremia (CHM) through novel study approaches.

> Dr. Kalatzis' research will deepen our knowledge about why damaged vital retinal proteins cause CHM."
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> *Neal Bench, CRF board president*

In Dr. Kalatzis' research, Unravelling the Pathophysiology of CHM using Innovative Approaches, her team will study how the absence of REP1 and the resulting prenylation defect give rise to the disease phenotype. Preliminary data suggest that this may be due to a dysregulation of ion channels, namely Ltype Ca2+. These channels regulate phagocytosis and growth factor secretion in the retinal pigment epithelium (RPE). Using live Ca2+ imaging, Dr. Kalatzis will comprehensively analyze the expression and activity of Ca2+ and Ca2+-dependent channels in iPSC-derived RPE generated from patients carrying diverse CHM mutations. The data will be further validated using specific channel inhibitors.

In parallel, an RNAseq study of the same panel of CHM iPSC-derived RPE will analyze this data to consolidate preliminary observations and identify novel pathways that may be dysregulated in CHM. Furthermore, the team transduced the same panel of RPE with an AAV vector expressing CHM and then applied RNAseq analysis to compare these transduced CHM lines with nontransduced and wild type lines. This approach will help validate candidate pathways and determine whether CHM replacement fully restores cellular activity.

"Dr. Kalatzis' research will deepen our knowledge about why damaged vital retinal proteins cause

CHM," 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."


Dr. Vasiliki Kalatzis with Institute for Neurosciences in Montpellier, France

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 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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