CHOROIDEREMIA RESEARCH FOUNDATION SUPPORTS RESEARCH INTO FEMALE CARRIERS OF A RARE INHERITED RETINAL DISEASE

Ms. Gocuk studies the first longitudinal assessment of progression rates in phenotype categories

SPRINGFIELD, MA, UNITED STATES, June 13, 2022 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce its latest research award to Sena A. Gocuk, BSci (Hons), D-OPTOM, MPhil, a PhD candidate at The University of Melbourne. Her research will expand the emerging science on female carriers of choroideremia (CHM), which has been largely unexplored.

> We are very excited to support Ms. Gocuk's work to expand the scientific community's knowledge about the unknown effects of CHM on female carriers."
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> *Neal Bench, CRF board president*

Choroideremia is an X-linked inherited retinal degeneration, caused by the disease-causing gene, CHM, resulting in nyctalopia and peripheral vision loss. Female carriers of CHM may present with variable retinal characteristics depending on expression of the CHM gene. In Ms. Gocuk's research, Structural and functional changes in female carriers of Choroideremia: A longitudinal study, she aims to re-examine the participants from the original study by Edwards et al (2015) that classified the different retinal phenotypes based on fundus autofluorescence (FAF) and other imaging modalities. Ms. Gocuk will obtain the first longitudinal assessment of female carriers of CHM with the hypothesis that there will have been different rates of progression in the phenotypic categories defined by Edwards. Using advanced multimodal imaging, Ms. Gocuk expects to learn more about the structural and functional changes in carriers over time.

Ms. Gocuk is the first recipient of an award from The Peter G. Boone Fund, the first endowment fund of the CRF announced in April 2021. Given by the Porter & Boone families, awards are exclusively designated to support early career investigators who are working towards, or have recently moved to, fully independent positions as investigators, faculty members, clinician scientists or scientific team leaders.

"We are very excited to support Ms. Gocuk's work to expand the scientific community's

knowledge about the unknown effects of CHM on female carriers," said Neal Bench, CRF board president.

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

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Sena Gocuk

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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