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

Research will identify the biomarker to predict disease progression

SPRINGFIELD, MA, UNITED STATES, February 15, 2023 /EINPresswire.com/ -- The Choroideremia Research Foundation (CRF) is pleased to announce the recipient of The Benelli Family 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) that she began in 2022 through a separate grant by the CRF. The award amount is $48,512.

> 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 expanded research, Advanced Genomic Analysis of Female Carriers of X-linked Retinal Disease, her team will use Nanopore long read sequencing and single cell RNA sequencing to determine the ratio and direction of lyonization more accurately in female CHM carriers. Lyonization is the process whereby only one of the two X-chromosomes (XC) is expressed in a particular cell. Depending on if a particular cell translates the wild-type or mutant gene, the spread of diseased cells in the retina will vary. As a result, the research may lead to identification of a biomarker that can be used to predict disease progression and may also be useful to determine who is eligible for emerging CHM therapies (including gene therapy).

"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.

Like males affected by CHM, female carriers may also present with clinical signs and symptoms much later in adulthood. Therefore, at the earlier stages, removing uncertainty regarding female carriers' final CHM severity and providing early forecasting of their prognosis are valuable for these women.


Sena Gocuk, recipient of The Benelli Family Research Award

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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