

Valkenburg, D. (2022). DISEASE SEVERITY IN FEMALE CARRIERS OF CHOROIDEREMIA AND A POSSIBLE ASSOCIATION WITH CHM EXPRESSION LEVELS.

*Inherited Retinal Diseases, 157.*

Conclusions of this research: Female carriers of choroideremia without exception (N=56) develop a disease phenotype ranging from mild retinal changes to extensive retinal degeneration, visual field loss and decreased BCVA in a substantial number of patients.

Reduced levels of wild-type CHM RNA expression, albeit measured in blood, may be associated with phenotype severity. The severity of disease encountered in this cohort supports the use of future gene(tic) therapies in at least a subgroup of patients.

**Link to relevant chapter in paper, Chapter 7:**

[LINK TO PAPER](#)