

Researchers find cells linked to leading cause of blindness in elderly

October 24 2019, by Bill Hathaway



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Age-related macular degeneration is one of the leading causes of blindness in the elderly, affecting more than 2 million people in the United States and leading to progressive loss of central vision. Genome wide studies have identified almost three dozen genes that play a role in the disease, but exactly where in the eye they inflict damage was not well



known.

Researchers from Yale University, the Broad Institute of the Massachusetts Institute of Technology, and Harvard University report in the Oct. 25 issue of the journal *Nature Communications* that glial cells (or support cells), and vasculature cells tasked with providing blood to the retina as well as cone cells contribute to degeneration of the macula, in the central part of the retina.

"This study helps pinpoint cell types that can be investigated closely to develop new types of therapeutics," said co-senior author Brian Hafler, assistant professor of ophthalmology and visual science and of pathology at Yale.

There are a limited number of effective long-term treatments available for the two forms of macular degeneration. The wet form is caused by growth of abnormal blood vessels underneath the macula, which can be mitigated by regular injections in the eye. Other than eye vitamin supplements, there is no treatment for the dry form of the disease, which is marked by accumulations of yellow deposits called drusen in the macula. While current treatments provide some benefits, over time there can be a continued, progressive loss of vision in both forms of the disease.

While genes associated with the risk of developing macular degeneration had been identified, the Yale/Harvard/MIT team used new single-cell sequencing to generate the first comprehensive human retinal atlas and employed data analysis technology to localize their effects to specific cell types associated with the disease.

While they found risk genes associated with cones, the cell type key to central vision, the researchers also found an association with glial and vascular <u>cells</u>—providing possible targets for novel therapies to improve



and restore vision.

Provided by Yale University

Citation: Researchers find cells linked to leading cause of blindness in elderly (2019, October 24) retrieved 6 May 2024 from https://medicalxpress.com/news/2019-10-cells-linked-elderly.html

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