

# Stem cell research reveals detailed genetic roadmap of glaucoma

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Acute angle closure glaucoma of the right eye (intraocular pressure was 42 in the right eye). Credit: James Heilman, MD/Wikipedia

A new, detailed genetic roadmap of glaucoma—the world's leading cause of irreversible blindness—will help researchers develop new drugs to combat the disease, by identifying potential target areas to stall or reverse vision loss.

The research, one of the largest and most detailed stem cell modeling studies reported for any disease, is published today in *Cell Genomics*.

By comparing stem cell models of the [retinal ganglion cells](#) of people with Primary Open Angle Glaucoma to those without the disease, more than 300 novel genetic features of these cells were uncovered.

The findings are the result of a national collaboration led by Professor Alex Hewitt (Centre for Eye Research Australia, University of Melbourne and University of Tasmania), Professor Alice Pébay and Dr. Maciej Daniszewski (University of Melbourne) and Ms Anne Senabouth and Professor Joseph Powell (Garvan Institute of Medical Research).

Professor Hewitt, who is Head of Clinical Genetics at CERA, says the study will lead to a better understanding of the mechanisms that damage retinal ganglion cells and lead to the onset of glaucoma.

This will help researchers develop [new drugs](#) to combat glaucoma, by identifying potential new areas to target to stall or reverse [vision loss](#) caused by the disease.

Healthy retinal ganglion cells—which transmit [visual information](#) from the eye to the brain via the optic nerve—are essential for [vision](#). In glaucoma, the gradual damage and death of these cells leads to a progressive, irreversible decline in sight.

"Glaucoma is often an inherited condition and comparing diseased retinal ganglion cells with healthy one is an effective way to increase our understanding of the mechanisms that contribute to vision loss," says Professor Hewitt.

Professor Pébay, whose team lead the stem cell aspects of this work, says that "until recently that's been impossible because you cannot obtain or profile retinal ganglion cells from living donors without an invasive procedure."

To overcome this challenge, the scientists used Nobel Prized-winning induced pluripotent stem cell (iPSC) technology to "reprogram" skin cells provided by donors into a stem cells that were then turned into a retinal ganglion cell in the lab.

They then mapped the individual genetic expression of almost a quarter of a million cells to identify features that could impact on the way genes are expressed in the cell, impacting its function, and potentially contributing to vision loss.

The researchers identified 312 unique genetic features in the retinal ganglion cell models that warrant further investigation.

"The sequencing identifies which genes are turned on in a cell, their level of activation and where they are turned on and off—like a road network with traffic lights," says Professor Powell, whose team led the analysis of world leading dataset.

"This research gives us a genetic roadmap of glaucoma and identifies 312 sites in the genome where these lights are blinking."

"Understanding which of these [traffic lights](#) should be turned off or on will be the next step in developing new therapies to prevent glaucoma."

Professor Hewitt, an ophthalmologist, says the research provides hundreds of new targets for researchers developing new drugs to treat [glaucoma](#) which is predicted to affect more than 80 million people globally by 2040.

"Current therapies are limited to slowing vision loss by reducing pressure in the eye—but they do not work for all patients and some people continue to lose many retinal ganglion [cells](#) and vision despite having normal eye pressure."

"The rich source of genetic information generated by this research is an important first step towards developing new treatments that go beyond lowering eye pressure and can reverse damage and vision loss."

**More information:** Alex W. Hewitt, Retinal ganglion cell-specific genetic regulation in primary open angle glaucoma, *Cell Genomics* (2022). DOI: 10.1016/j.xgen.2022.100142. [www.cell.com/cell-genomics/ful...2666-979X\(22\)00075-1](http://www.cell.com/cell-genomics/ful...2666-979X(22)00075-1)

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