Gene study sheds light on middle-age sight loss

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Chemical changes in the eye that can lead to blindness have been identified by scientists, a conference will hear tomorrow (Tuesday 5 September).

Their findings aid understanding of a genetic condition that causes sight loss for one in 3,000 people in the UK.

They will be presented at the Eye Development and Degeneration 2017 conference in Edinburgh.

Eye condition

Scientists examined how changes in a gene known as RPGR damage eye cells to cause a disorder known as X-linked retinitis pigmentosa.

The condition is incurable and affects night and peripheral vision before gradually causing blindness in middle age.

Skin samples

A team led by Edinburgh researchers took skin samples from two patients and transformed stem cells – which can change into any cell type – into light-sensing eye cells known as photoreceptors.

They compared these with cells from healthy relatives of the patients.

Photoreceptors – which decay in retinitis pigmentosa patients – differed in their fundamental structure when compared with those from family members.

Key molecules

Follow-up studies in mice identified key molecules that interact with RPGR to maintain the structure of photoreceptors.

When RPGR is flawed, the structure is compromised and photoreceptors cannot function correctly, leading to sight loss.

The study is published in the journal *Nature Communications* and was carried out at the University of Edinburgh's Medical Research Council Centre for Regenerative Medicine.

It was funded by the Wellcome Trust and the charity Retinitis Pigmentosa Fighting Blindness.

"By furthering our understanding of the RPGR gene and its effects on photoreceptor cells, we hope our findings bring us closer to developing a possible treatment for this devastating disease," says Dr Roly Megaw of the Medical Research Council Institute for Genetics and Molecular Medicine.

Provided by University of Edinburgh