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Breakthrough in identifying new genes in age-related macular degeneration, the leading cause of blindness

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Fight for Sight funded research published this week in *Clinical Epigenetics*, has identified new genes linked to the development of age-related macular degeneration, the leading cause of blindness in the UK.

The research team, led by Dr. Louise Porter at the University of Liverpool, have identified genes that may represent new targets for the development of treatments. Currently there is no available treatment for 85% of people with the common 'dry' from of the disease.

Researchers used cells from 44 human donor eyes to profile the levels of DNA methylation – a chemical change that may be influenced by sex, age, smoking and diet—and looked at the underlying gene changes in age-related macular degeneration. In doing so they were able to identify changes in specific genes that were not previously known to be linked to the condition.

The team's finding paves the way for testing new treatments that could target the affected genes.

Dr. Neil Ebenezer, Director of Research, Policy and Innovation at Fight for Sight, said: "There is currently no treatment for dry age-related macular degeneration so the results from this study are extremely positive and bring hope for people living with this condition. By identifying new gene targets, researchers have more options for

developing new treatments."

Fight for Sight funded researcher and lead author, Dr. Louise Porter, from the University of Liverpool, said: "Our main aim for conducting this research was to help tackle an area of unmet clinical need. This work has identified new genes, providing us with novel targets for investigation in a disease in desperate need for therapies."

Age-related macular degeneration is the leading cause of blindness in the UK and currently affects 600,000 people. The condition affects the macula which is a small part of the light sensitive layer at the back of the eye known as the retina. The condition causes blurred or reduced central vision in one or both eyes. The disease significantly affects quality of life, making it difficult to see details, like recognise people's faces and read. There is currently no treatment for patients living with the most common form of macular degeneration, known as 'dry' macular degeneration.

Previous research has been carried out to understand the genetic cause of the disease. However, it is still unknown what causes the development of macular degeneration.

The researchers observed that some genes with different levels of methylation also had varying amounts of gene product – (RNA) - expressed, suggesting a role for DNA methylation in abnormal gene regulation in the disease.

More information: Louise F. Porter et al. Whole-genome methylation profiling of the retinal pigment epithelium of individuals with age-related macular degeneration reveals differential methylation of the SKI, GTF2H4, and TNXB genes, *Clinical Epigenetics* (2019). DOI: 10.1186/s13148-019-0608-2

Provided by Fight for Sight