New method to improve diagnostics and treatment for corneal dystrophy

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The results from a Fight for Sight funded study pave the way for early

diagnosis of a corneal eye condition, as well as aiding the development of new targeted treatments.

The study published in *Genetics in Medicine* last week used an innovative method to sequence the gene mutation responsible for more than three quarters of cases of an eye condition called Fuchs Endothelial Corneal Dystrophy (FECD). The mutated region has not previously been sequenced accurately.

Researchers led by Dr. Alice Davidson at UCL applied a new technique known as 'No Amp Targeted Sequencing' which enables the mutation to be accurately sequenced, for the first time in patient samples.

The sequencing technique will help the development of new treatments targeted to the mutation. The technique could also detect the condition before symptoms have begun to show, which will help identify patients suitable for preventative treatments.

Results from this study could also have wider implications for more than 40 human diseases such as Huntington's disease, which are caused by a similar type of mutation.

Dr. Neil Ebenezer, Director of Research, Policy and Innovation at Fight for Sight, said: "Currently some gene sequences are very difficult to faithfully recreate so this new technique means researchers will be able to more accurately recreate gene sequences, improve diagnosis and help develop treatments.

He added "It's particularly exciting as this technique also has potential applications for more than forty other conditions."

Dr. Alice Davidson, a Fight for Sight funded researcher from UCL Institute of Ophthalmology, said: "I am enormously excited by our

application of this innovative method. The technique has already improved our understanding of Fuchs endothelial corneal dystrophy biology and hopefully, in the future, will help facilitate more effective diagnosis and treatment for patients."

Fuchs Dystrophy is an inherited, sight threatening condition which is estimated to affect up to 4.5% of individuals over 50 years of age. The condition affects the cornea, causing light sensitivity, cloudy vision and the ability to see details clearly.

More information: Hafford-Tear et al. CRISPR/Cas9-targeted enrichment and long-read sequencing of the Fuchs endothelial corneal dystrophy–associated TCF4 triplet repeat. *Genetics in Medicine*. 2019. doi.org/10.1038/s41436-019-0453-x

Provided by Fight for Sight