Genetic factors behind eye disorder identified

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(Medical Xpress)—Researchers at the University of Liverpool have identified genetic changes in a gene found in more than 12% of patients with a degenerative eye disorder.

Professor Colin Willoughby, from the Institute of Ageing and Chronic Disease, has shown that a genetic change, termed ZNF469, could provide valuable information into how patients develop a condition called keratoconus.

Keratoconus is a life-long condition affecting the cornea, the clear covering of the eye, making the cornea become a cone-like shape. This causes vision to become blurred and distorted.

Professor Willoughby said: "This is the most significant genetic factor responsible for keratoconus identified to date. Understanding the genetic basis of this common corneal condition in young adults will provide insight into why keratoconus develops and could ultimately lead to new therapies.

Corneal thickness

"The ZNF469 gene is also involved in corneal thickness which is a key risk factor for glaucoma; the leading cause of irreversible blindness affecting more than 60 million people worldwide."

Although spectacles or contact lenses usually help people with less severe keratoconus, sometimes surgery is required. Eventually, some patients may need a corneal transplant. The procedure, however, carries risks and transplants are not always successful.

The underlying biochemical changes and genetic defects of keratoconus are poorly understood, but this new research signals new hope for patients with the condition. It is thought that new treatments such as collagen cross-linking may prevent progression of the disease in some patients, but an understanding of its genetic base could help prevent the disease on a larger scale.

The research conducted by an international consortium of researchers has been published in *Human Molecular Genetics*.

Provided by University of Liverpool