20 year-old puzzle solved through genetic advances

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Members of the Ridland and Kadiri family with Prof Colin Willoughby (Back row far left), Dr Vito Romano (Back row second from left) and Dr Kevin Hamill (Back row far right)

Researchers from the University of Liverpool have identified a specific

gene that plays a key role in an inherited eye disorder.

The discovery solves a 20 year-old puzzle for a family in Liverpool who all developed epithelial recurrent erosion dystrophy (ERED), a genetic corneal dystrophy disorder that causes abnormality of the outer layer of the eye.

Corneal dystrophies may not cause symptoms in some individuals; in others they may cause significant vision impairment. These disorders are caused by many different genes some of which are known but many have yet to be identified.

The Leaving of Liverpool

The Liverpool family, Ridland and Kadiri, first met Professor Colin Willoughby, from the University's Institute of Ageing and Chronic Disease (IACD), when he was a junior doctor and corneal fellow at St. Paul's Eye Unit in 1996. They had a type of corneal dystrophy, as yet unidentified, which caused spontaneous, recurrent, painful scratches on the cornea from early life.

In discussion with New Zealand-based Opthalmologist, Dr Andrea Vincent, Professor Willoughby learnt that there were similar families in New Zealand and Australia. Working together they sought to characterise the gene responsible for this particular disorder by using genomics.

Professor Willoughby, said: "Technological advances in the clinic and laboratory have allowed us a greater appreciation of the observable physical or biochemical characteristics of an organism and the genetic diversity of corneal dystrophies.

"As a result of this study we are now able to offer a genetic diagnosis of

the disease before the actual symptoms of the disease have appeared. We can also give advice to parents concerning the risks associated with this disorder in a child."

Greater Understanding

Epithelial recurrent erosion dystrophy (ERED) is a rare form of superficial corneal dystrophy which affects the clear window on the front of the eye. Patients develop recurrent, painful scratches on the cornea damaging its outermost layer and healing with a scar. These erosions which usually commence in childhood are painful and lead to intense light sensitivity and reduced vision.

Laser treatment can help alleviate the symptoms and although this does not cure the disease it does slow it down. No treatments are available to correct the genetic defect at the present time.

Lecturer in Cell and Molecular Biology at the University's Department of Eye and Vision Science (IACD), Dr Kevin Hamill, said: "The protein identified in this study is called collagen 17 and it has a pivotal role in the skin. And, from this study, the eye. Understanding the function of these types of proteins have broader implications for wound healing in the body and scarring in the eye.

"This work shows the power of genomics and success of the unique research environment here at the Institute of Ageing and Chronic Disease for studying the genetics and biology of eye disease."

More information: Verity F. Oliver et al. A COL17A1 Splice-Altering Mutation Is Prevalent in Inherited Recurrent Corneal Erosions, *Ophthalmology* (2016). DOI: 10.1016/j.ophtha.2015.12.008

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