New finding in rare eye disease

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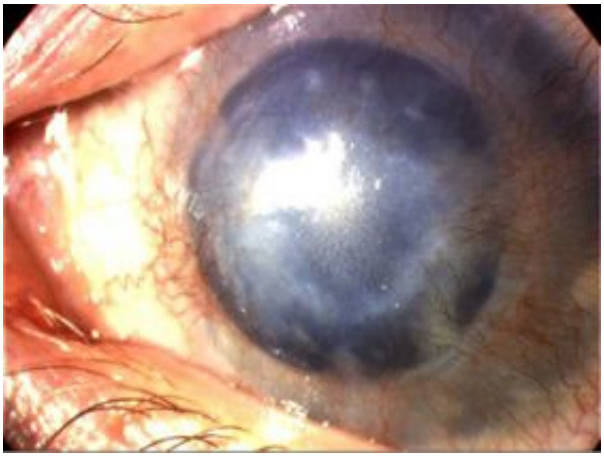

(PhysOrg.com) -- Researchers have made a finding which could pave the way for new treatments for sufferers of a rare eye disease which can lead to blindness.

In a paper published in the *Journal of Pathology*, scientists reveal they have discovered why mutations in a key gene can cause the cornea to go opaque and lead to sight loss.

Pax6 is the gene responsible for the development of the eye and mutations of it can cause the cornea to go cloudy.

Corneas - our windows on the world – are delicate, transparent tissues which are constantly exposed to potentially damaging environmental

factors such as daylight, ultraviolet light and high levels of oxygen.

Normal corneas contain proteins that protect against the day to day stress of coping with these environmental stresses.

But around one in 40,000 people have Pax6 mutations which lead to them developing a condition called Aniridia, where the iris of the eye is missing.

As well as having no iris, most sufferers will develop some degree of corneal cloudyness which is called Aniridia-Related Keratopathy or ARK.

Now researchers have found that people with ARK have tiny holes in their corneas, which open up the corneas to extra levels of stress. They also discovered that they have less of the protective proteins, which results in their eyes being unable to cope with everyday conditions. Over time, this leads to the corneas going cloudy.

Dr Martin Collinson, a Senior Lecturer in Biomedical Sciences at the University of Aberdeen, led the research team. He said: "In the worst cases Aniridia-Related Keratopathy can be chronically painful and sight threatening. Current therapy is surgery, anti-inflammatory drugs, or nothing at all."

The researchers found that if they applied chemicals to diseased corneas, they were able to reduce the stress caused by oxygen. This in turn gave cells on the eye's surface more of a fighting chance of protecting the eye.

Dr Collinson added: "Our findings provide answers to a disease that has been very poorly understood and is rather difficult to treat.

"We hope that our studies could potentially lead to new treatments for

people with ARK that could involve eye drops as opposed to surgery."

Source: University of Aberdeen