'Lazy eye' discovery of how an old gene learns new tricks

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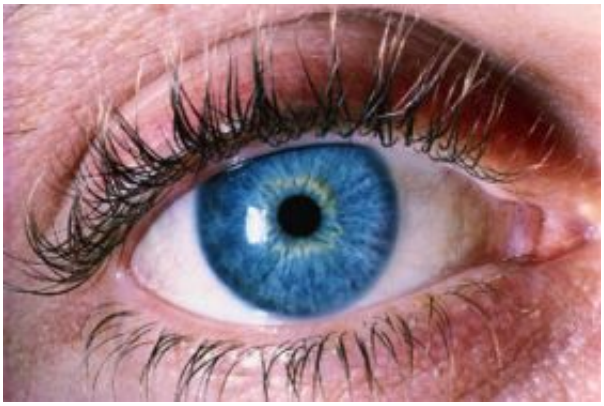

(PhysOrg.com) -- Researchers have made a discovery which could lead the way for new treatments into a rare eye disorder which if not treated can result in permanent blindness in childhood.

An eye disorder which leads to "lazy eye" (strabismus) first described in early 1900, and a gene known since 1990 to be widely expressed within the nervous system, have now been linked together.

Mutations of the CHN1 gene give rise to a hyperactive gene product called a2 chimerin that in turn affects the normal eye development.

These new findings published in *Science* today demonstrate how genetic

errors can explain developmental errors.

The research shows that seven CHN1 mutations found in families with a history of Duane's Retraction Syndrome (DRS) - a rare, disorder of eye movement which is present at birth - lead to abnormal development of the cranial nerve III which is integral to normal eye development.

A large team of experts from UK and USA, including a scientist now based in Aberdeen, have been involved in the research into the CHN1 gene and its connections to this unique syndrome which affects 1% of the general population of individuals with eye movement disorders worldwide. The condition may affect one or both eyes and is more common in girls.

Dr Maria Psatha, University of Aberdeen who co-authored the paper said: "In normal eye movements, 3 cranial nerves control 6 eye muscles, which control the movement of each eye horizontally, up and down, or at an angle. In DRS,miswiring between the muscles and the nerves can cause some eye muscles to contract when they should not and other eye muscles not to contract when they should. This typically occurs around the sixth week of pregnancy when the cranial nerves and eye muscles develop. During this time, the mutations of the gene of CHN1 have now been shown to explain the pathophysiology of the DRS disorder."

It is hoped that this latest discovery will lead to future developments in the treatment of Duane's Syndrome.

Dr Psatha,continues: "I have been very fortunate to be part of a very fruitful international interdisciplinary collaboration that resulted in a high impact paper reporting the findings of the gene responsible for an important yet rare eye disorder that affects children. I look forward to more collaboration of this calibre in the very vibrant environment of the University of Aberdeen."