Researchers identify gene mutation that can cause key-hole shape defect in eye

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A scientific collaboration, researchers from The University of Manchester at the Manchester Centre for Genomic Medicine (MCGM), and the Telethon Institute of Genetics and Medicine (TIGEM) in Naples, , has pinpointed the genetic cause of a rare form of blindness, which can present itself as a key-hole shaped defect in the eye in newborn babies.

Professor Graeme Black

The condition is known as inherited retinal dystrophy associated with ocular coloboma.

Coloboma is one of a number of developmental genetic disorders that collectively represent important causes of visual disability affecting one in 4000 people in the western world. Results of this study, which was funded in the UK by charities Fight for Sight andRP Fighting Blindness, could help scientists better understand the link between genetics and disease, and speed up the rate at which patients with this group of conditions are diagnosed. The work also provides fundamental insights into the earliest genes that are required to control the development of the eye.

This European research consortium identified a mutation in the miR-204 gene as being responsible for the condition, and the findings were published in the
PNAS journal.

Patients with coloboma are born with a hole in one of the structures of their eye, such as the iris or retina, which fails to close up. In this study, researchers investigated instances where the condition affected both eyes and was associated with progressive visual loss from a degeneration of the light sensitive cells of the eye, the photoreceptors.

One of the two principal investigators, Professor Graeme Black, who is also Strategic Director at the MCGM and Consultant in Genetics and Ophthalmology said: "Around 200 genes have previously been linked to inherited developmental and degenerative genetic disorders. For the first time, we've been able to demonstrate the importance of the miR-204 gene in the regulation of ocular development and maintenance, and of its contribution to eye disease.

"This discovery provides a clearer understanding of the control of early eye development as well and helping to improve diagnosis for patients with this condition and possibly other inherited eye disorders."

Dr Sandro Banfi, co-principal investigator of this work, who leads a research group at the Telethon Institute of Genetics and Medicine and is Professor of Medical Genetics at the Second University of Naples, said: "This is an extraordinary result that sheds further light on the role of microRNAs, very tiny genes that have been recently discovered, as primary causes of genetic diseases. This success would not have been possible without the collaboration between our two research groups.

"It's very exciting to see such progress being made in the project," commented Sue Drew, Engagement Manager at RP Fighting Blindness. "We're delighted to see scientific collaboration delivering real results for retinal dystrophy research which has increased understanding of genetic disease, and will aid diagnosis in the future. We anticipate further exciting developments being made and hope this project will show the huge benefits of collaborative research."

Founded by Central Manchester University Hospitals and The University of Manchester, MCGM is an integrated academic and clinical service department. The Telethon Institute of Genetics and Medicine, founded by the ItalianTelethon Foundation, is a research institute dedicated to the study of mechanisms underlying genetic diseases and to the development of strategies for prevention and treatment.

The study investigated the genetic cause of an autosomal dominant inherited condition of retinal dystrophy and bilateral coloboma, present in varying degrees in a large, five-generation family.

Conte I, Hadfield KD, Barbato S, Carrella S, Pizzo M, Bhat RS, Carissimo A, Karali M, Porter LF, Urquhart J, Hateley S, O'Sullivan J, Manson FD, Neuhauss SC, Banfi S, Black GC. MiR-204 is responsible for inherited retinal dystrophy associated with ocular coloboma. Proc Natl Acad Sci U S A. 2015 Jun 8. pii: 201401464. [Epub ahead of print]

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