

Myopia and glaucoma genes identified

By <u>Dr Ananya Mandal, MD</u>

In global studies of more than 40 000 people from countries around the world, including Australia, researchers have found the genes responsible for two of the most common eye disorders - short-sightedness or myopia and glaucoma. The scientists believe this could open new diagnostic and management options.

Stuart MacGregor, of the Queensland Institute of Medical Research, who was on the research team, said about one-third of Australians have some degree of shortsightedness, or myopia, although the condition was more common in Asian people. He added, "Myopia is also a risk factor for a range of other eye diseases." The gene is the first to be found that is linked to the common form of the condition in Caucasians. He added that this discovery could pave the way for gene therapy for myopia in future. The increase in occurrence of myopia during the past century was mainly due to environmental factors such as doing close work and reading, and there is some evidence that spending time outdoors and looking into the distance could help prevent it, he said.

Finding the markers

Professor David Mackey, an ophthalmologist at the Lions Eye Institute in Western Australia, was part of the project to identify the exact genes responsible for myopia. "We analyse usually around 600 000 DNA markers and find which markers tend to run more commonly with the feature that we are measuring - in this case myopia...And in collaboration with the twin research group in London we have been able to identify one new gene associated with myopia, mainly in older people," he said. He explained that three million Australians suffer from myopia but the real epidemic is across Asia.

"Particularly Singapore, Taiwan, Hong Kong and now the large developing cities in China, [where] a majority of children when they finish high school are myopic, needing to wear glasses... Now this is something that has happened really in the last 50 years and we are not sure what it is that led to this epidemic but we suspect that the east Asians are more genetically predisposed to getting myopia... Therefore understanding the underlying mechanism is of great importance, particularly if we are not having to supply glasses to the entire population of east Asia."

A drug could be developed

Speaking on the potential of this study Dr. Mackey said, "One of these particular genes involved in the risk of myopia is something that we can actually develop a drug to intervene for, or we may be able to come up with other treatments, such as whether people should or shouldn't wear glasses all the time, whether they should get outdoors, or how much reading should they be doing... All of these are factors that have been proposed as risk factors for developing myopia in those who are predisposed to get it."

Dr MacGregor explained that decreased thickness of the cornea is one of the main causes of glaucoma, a chronic degenerative condition that leads to increased pressure in the eyes and a leading cause of irreversible blindness worldwide. "Our research is the first to identify a gene that influences corneal thickness," he said. He said that genes also influence the pressure in the eye.

According to Dr MacGregor many more genes were likely to be linked to the two conditions. A gene linked to a leading cause of blindness in children, called optic nerve hypoplasia, was also identified.

The study was a part of an international project in collaboration with a team at the Centre for Eye Research Australia led by Professor David Mackey and is published in the journal *Nature Genetics*.

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