

# Genes appear to play significant role in the development of cortical cataract

**Devon Schuyler**

Genetic factors appear to affect the risk of cortical cataract, but not posterior subcapsular cataract (PSC), according to a recent Johns Hopkins study. The finding could help in developing strategies for prevention of cortical cataract.

"Many previous studies have concentrated on nuclear cataracts, which are the most common age-related cataracts in the Western world. However, cortical cataracts are the most common age-related cataracts in people of Indian and African descent, and this work provides them with needed attention," J. Fielding Hejtmanck MD, of the ophthalmic genetics and visual function branch of the National Eye Institute, told *EuroTimes*.

The study included 321 participants (average age, 78) from the Salisbury Eye Evaluation study, an ongoing study of older white and black people living on Maryland's Eastern Shore, and 453 of their siblings (average age, 73). Nearly 30% of the participants were black.

The researchers determined the presence or absence of cataract by reviewing existing photographs and clinical grades or by taking

new digital retro-illumination photographs. Cortical cataract was defined as a grade of 4/16 or greater based on grading in units of 1/16. PSC was graded as either present, absent or questionable; only grades of "present" counted as cases.

After adjusting for factors associated with cataract risk, such as use of tobacco, alcohol, oestrogen and steroids; lifetime exposure to ultraviolet-B light; serum antioxidant levels; and the presence of medical conditions such as diabetes, researchers found that the magnitude of heritability was 24% for cortical cataract. The heritability of PSC was not statistically significant.

## Several risk factors identified

Factors that increased the odds of cortical cataract were increasing age, being female, having a history of diabetes, and being black. High blood levels of vitamin A were associated with a reduced risk of cortical cataract. Factors that increased the odds of PSC were a history of diabetes and use of topical or oral steroids. All of these findings were consistent with those of other published reports.

An unexpected finding was the lack of association between cataract and lifetime exposure to ultraviolet-B light; previous studies have found an association between the two. This inconsistency might be explained by the relatively small size of this study.

Prior to this study, a population study had suggested that cortical cataracts tended to cluster in families and a twin study had suggested that the magnitude of heritability for cortical cataracts might be about 58%.

The relatively low magnitude of heritability in this study might be explained by the fact that the average age of the participants was in the late 70s, compared with age 62 in the twin study. Older people have had more years of exposure to environmental factors such as ultraviolet light, reducing the relative importance of genes.

In addition to its relatively small size and older population age, possible weaknesses of this study include the use of old photographs and clinical grades to determine the presence of cataract, and the fact that less than half of the eligible siblings participated in the study.

The study authors pointed out that using an older population may also be advantageous because it reduces the risk of misclassifying younger individuals as unaffected by cataract when they will be developing cataract in the future.

## Prevention better than cure

Nathan Congdon MD MPH, the lead author of this study, is an associate professor of ophthalmology and international health at the Johns Hopkins University School of Medicine. He told *EuroTimes* that the impetus for this study came from his desire to find ways to prevent cataract, which is a growing problem worldwide because of the increasing age of the population.

"In the United States and other Western countries, it's relatively easy to do cataract surgery. But in poorer countries, access to surgery is limited by a lack of funds and trained surgeons. That's why trying to figure out how to prevent cataract is really critical in places like Africa and Asia. Having a way to prevent cataract rather than needing to operate on each case would be much more beneficial approach from a global standpoint."

Dr Congdon has been involved in attempts to develop medications to prevent cataract formation, all of which have been unsuccessful so far. He explained that learning more about the genetic basis of cataract could help researchers develop successful strategies.

Towards this end, the next stage of the Salisbury Eye Evaluation study will involve an attempt to locate genes that affect cataract formation. The Centre for Inherited Disease Research will be analysing blood samples from the study for single nucleotide polymorphisms (SNPs) to see which ones might relate to the different types of cataract.

"If we had a better understanding of what exactly the pathways are that lead to cataract from a protein standpoint, then we'd do a better job of coming up with interventions against cataract," he said.

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