New glaucoma research solves anthropological and medical puzzle

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Scientists studying a rare form of glaucoma have discovered why people in the disparate Roma communities are at greater risk of inheriting a condition leading to permanent blindness than other groups in the population.

Primary congenital glaucoma (PCG) is a devastating condition affecting 1 in every 1000 Romany people. Researchers at the University of Leeds, looking to uncover the cause, found a single gene mutation repeatedly appearing in affected families.

An international collaboration led by Dr Manir Ali of the Leeds Institute of Molecular Medicine, first identified the 'Jatt' mutation in one of four Pakistani families. Further study amongst Roma populations in Europe showed that the same mutation accounted for nearly half of all cases of PCG in that community.

Dr Ali's research also confirms the widely accepted view that the Roma originated from the Jatt clan of Northern India and Pakistan and not from Eastern Europe as previously believed.

Dr Ali said: "Glaucoma is the leading cause of preventable blindness worldwide and we are now looking at this same gene to see if changes in it are also found in patients with the more common form of glaucoma, primary open angle glaucoma (POAG) or chronic glaucoma, which affects so many older people.

“It is hoped that our research, which looked at a relatively rare form of the disease, can help medical professionals address the health needs and find appropriate treatments for a particularly vulnerable at-risk group,” he added.

Recognised as Europe’s largest ethnic minority, the Romany and Gypsy communities continue to suffer discrimination. An estimated 84% across Europe live below the recognised poverty line.

A recent Mori poll revealed that a third of UK residents admitted to being prejudiced against these groups. The British Medical Association says the Gypsy communities have the lowest life expectancy and the highest rate of child mortality in the UK.

One reason may be the difficulty these groups have in registering with a GP, which effectively excludes them from the health care system, routine screening and early diagnosis of disease.

Although the common form, chronic glaucoma, is difficult to detect in the early stages, early diagnosis offers real opportunities to treat this condition. However, if left unchecked it leads to loss of vision and in some cases permanent blindness.

This means that regular eye examinations are essential, especially in high-risk groups, such as the elderly and those with a family history of the condition.

More information: Null Mutations in LTBP2 Cause Primary Congenital Glaucoma, Manir Ali, Martin McKibbin, et al. *The American Journal of Human Genetics* - 09 April 2009, 10.1016/j.ajhg.2009.03.017

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