

# Leicester breakthrough in eye disease

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Researchers at the University of Leicester have identified for the first time a gene which causes a distressing eye condition. Their discovery, as reported in the journal *Nature Genetics*, is expected to lead to better treatments for the condition.

Nystagmus causes the eyes to move in an uncontrollable manner, so that people with the condition cannot keep their eyes still. Nystagmus can be congenital (occurs at birth or in early childhood) or acquired later in life due to neurological disease.

Congenital nystagmus is frequently genetic. Treatment can be surgical, by correcting an abnormal head position (which occurs because the nystagmus is quietest in a certain direction of gaze) or by correcting a squint. The effects of Nystagmus can also be reduced by drugs.

Recently, the University of Leicester Ophthalmology Group, headed by Professor Irene Gottlob, has shown that drug treatment is helpful in congenital nystagmus, as well as in the form that develops later.

The frequency of nystagmus is unknown. However, over the last six years the Leicester Ophthalmology Group has counted all patients with the condition in the 'Leicestershire Nystagmus Survey', showing an occurrence of more than two in 1000 people.

Professor Gottlob commented: "The discovery of this gene will make a genetic test for idiopathic X-linked nystagmus possible. So far it has not been understood what the causes of nystagmus are. The discovery of the

gene will lead to greater understanding about the protein which is abnormal in nystagmus.

"Our research also showed that the expression of the protein is changed in neuronal cells of the eye and in certain parts of the brain. Further research is now needed to understand what functional changes in the brain the gene mutations are causing.

"This will be the first time the mechanisms of nystagmus have been understood, and we hope it will lead to better drug treatments. Understanding the mechanism of nystagmus will also improve our knowledge of the control in eye movements in general."

Nystagmus, largely under-researched, is one of a significant number of interests within the Leicester Ophthalmology Group concerned with normal and abnormal eye movements. Researchers are looking into many aspects of nystagmus, including possible drug treatments, its epidemiology, impact on visual function, adaptation of the visual system to the constant eye movements, the causes of the condition and its genetic make-up.

They are also investigating other eye movement problems, such as reading in schizophrenia and treatment for amblyopia (a lazy eye), including patching therapy with special reward incentives and the education of parents and teachers.

Source: University of Leicester

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