New gene discovery could help to prevent blindness

11 February 2010

Scientists have uncovered a new gene that could help save the sight of patients with a type of inherited blindness.

The international research team led by the University of Leeds found that the TSPAN12 gene is faulty in patients with a disease known as FEVR (Familial Exudative Vitreoretinopathy), which affects the development of the eye.

While many FEVR patients are registered blind or visually impaired, members of the same family may carry the faulty gene without showing any symptoms. It is hoped that by screening these family members for TSPAN12 mutations, doctors may be able to catch FEVR early on and treat patients before they start to lose their sight. It will also broaden their understanding of other more common blinding disorders.

Dr Carmel Toomes, of the Leeds Institute of Molecular Medicine who led the research, said: "This discovery will have an immediate impact on the treatment and counselling of some FEVR patients by allowing us to identify family members who carry the mutated gene before any retinal damage has occurred. This decreases their chances of going blind because if a patient is diagnosed early enough their sight can often be saved by surgical intervention."

TSPAN12 is thought to cause FEVR by disrupting the cell signals required for the normal development of blood vessels in the retina at the back of the eye.

This study, which was funded by The Royal Society and the Wellcome Trust, looked at 70 FEVR patients who had tested negative for the three genes already known to cause the disease. Mutations in the TSPAN12 gene, which is located on chromosome 7, were found in 10% of these patients.

As well as being an important piece in the FEVR puzzle, this latest discovery will help scientists to understand other blinding disorders including age-related macular degeneration and diabetic retinopathy - two of the leading causes of blindness in the developed world.

"Our research highlights how studying rare inherited disorders such as FEVR can help us identify the genes and pathways involved in the basic cellular processes underlying more common diseases," Dr Toomes added.

"We hope that by learning more about blood vessel formation in FEVR we will gain clues that may lead to new treatments for these conditions."

The research will be published in the *American Journal of Human Genetics* on 12th February.

More information: Poulter JA et al. Mutations in TSPAN12 cause autosomal-dominant familial exudative vitreoretinopathy. Am J Hum Genet 2010;86:248-253.

Provided by University of Leeds