EU research sheds light on causes of blindness

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Over 15 million people in Europe are visually impaired, and 2.7 million are totally blind. The commonest cause of blindness, age-related macular degeneration (AMD) accounts for 12.5 million of these cases. However, there are also many thousands of people affected by rarer inherited forms of blindness.

Understanding the genetic causes of blindness is the overarching goal of the project. As the name implies, the project is focused in particular on the genes behind the retina, the structure which contains the photoreceptor cells responsible for converting light into nerve impulses.

EVI-GENORET hit the headlines two months ago when one of the project partners, Professor Robin Ali of the UK's Institute of Ophthalmology, announced the start of the world's first ever clinical trial to treat a rare form of inherited blindness with gene therapy.

The disease, Leber's congenital amaurosis (LCA), is caused by a mutation in a gene which controls the production of an enzyme which the eye needs to be able to capture light effectively. The new treatment entails injecting healthy versions of the gene into the retina, where they produce the enzyme correctly.

Trials in dogs proved successful and two human patients have since received the

treatment. In total, Professor Ali and his team plan to test the new treatment in 12 patients. So far there have been no complications, and the team running the trial hope to be able to present the first results in a year's time. Meanwhile, Professor Ali is confident about the role gene therapy is set to play in treating blindness in the future.

'It is my prediction that within five years, gene therapy will be an effective treatment of choice for a number of inherited retinal disorders,' he stated.

Another important success of the project is the establishment of the EVI-GENORET database. 'This doesn't sound exciting but it is crucial to the success of the project,' said project coordinator Professor José-Alain Sahel of France's Institute of Health and Medical Research.

The database brings together in a standardised way all the diverse information produced by the project partners. This includes samples, animals, cells, images and data. In the future the partners hope to expand the database to cover other countries from around the world.

In addition to the LCA gene, the project partners have succeeded in identifying a number of other genes which contribute to the correct functioning of the retina. They are particularly keen to identify the factors which cause AMD.

A particular characteristic of the project is the close involvement of patients in the work. One of the project partners is Retina International (RI), an umbrella organisation which promotes research into degenerative retinal diseases and provides support to people affected by these diseases. RI's members spend €30 million on research annually, Christina Fasser of RI told CORDIS News.

Ms Fasser has retinitis pigmentosa, a genetic disease in which vision gradually deteriorates over a number of years. She is now completely blind, and for her the motivation for taking part in research projects such as EVI-GENORET is simple. 'It gives us hope,' she said.

EVI-GENORET brings together over 24 organisations, including academic institutions, businesses, a management company and a patient group. The four year project has received €10 million in funding from the Sixth Framework Programme's 'Life sciences, genomics and biotechnology for health' thematic area.

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