First clinical trial of gene therapy for childhood blindness

May 1 2007

The first clinical trial to test a revolutionary treatment for blindness in children has been announced by researchers at UCL (University College London). The trial, funded by the Department of Health, is the first of its kind and could have a significant impact on future treatments for eye disease.

The trial involves adults and children who have a condition called Leber's congenital amaurosis (LCA), which is a type of inherited retinal degeneration. This disease causes progressive deterioration in vision, due to an abnormality in a particular gene called RPE65. This defect prevents normal function of the retina, the light-sensitive layer of cells at the back of the eye. This results in severely impaired vision from a very young age and there are currently no effective treatments available.

The new technique that will be used in the trial involves inserting healthy copies of the gene into the cells of the retina to help them to function normally. Restoring the activity in these cells should restore vision. The operation delivers the normal genes to the retina, using a harmless virus or "vector" to carry the gene into the cells. The vector has been manufactured for this trial by Targeted Genetics, Seattle, USA.

Previous work using animal models has demonstrated that this gene therapy can improve and preserve vision. During trials, the vision of dogs with the defect was restored to the extent that they were able to walk through a maze without difficulty; something they could not do before the treatment. As this trial is the first to treat an eye disease using

administration of gene therapy vectors to human retinas, the team have carried out extensive pre-clinical testing. The purpose of this trial is to find out how safe and effective the new intervention is in humans.

The team conducting the trial, from UCL Institute of Ophthalmology and Moorfields Eye Hospital, is led by Professor Robin Ali and includes leading eye surgeon Mr James Bainbridge and leading retinal specialist Professor Tony Moore.

Professor Ali said: "We have been developing gene therapy for eye disease for almost 15 years but until now we have been evaluating the technology only in the laboratory. Testing it for the first time in patients is very important and exciting, and represents a huge step towards establishing gene therapy for the treatment of many different eye conditions."

The trial's first operations have already taken place in young adult patients who developed the condition as children. Mr James Bainbridge, who leads the surgical team, said: "It is very encouraging that we can deliver genes to an extremely fragile site in the eye without complications."

Professor Moore said: "Some indications of the results of the trial may be available within several months. However, the subjects will need to be followed-up to assess the long term effect of the treatment. It will be many months before we have the full picture. We anticipate the best outcome in younger patients, as we will be treating the disease in the early stages of its development."

Professor Ali added: "There are many forms of retinal degeneration, meaning the use of gene therapy treatments must be individually developed then tested in a separate clinical trial specifically for that disease. However, the results from this first human trial are likely to

provide an important basis for many more gene therapy protocols in the future, as well as potentially leading to an effective treatment for a rare but debilitating disease."

Source: University College London