New gene therapy treatment boosts quest for vision loss cures

Oxford University researchers hope study will pave way for single-treatment cure for many types of inherited blindness

 Prof Robert MacLaren: ‘As we learn more about genetics, we realise that correcting faulty genes even before a disease starts may be the most effective treatment.’ Photograph: University of Oxford/PA

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Groundbreaking gene therapy has restored some vision to patients who were going blind.

[University of Oxford](http://www.theguardian.com/education/oxforduniversity) researchers hope the findings of the small study will lead to potential cures for common causes of vision loss, including genetic-related macular degeneration, which affects thousands of people in the UK.

The results showed some people had improvement in vision for up to four years. They had been suffering from choroideremia, which is caused by a defect in the gene CHM and affects about one in 50,000 people in the UK. Currently there is no cure or treatment for the disease, which progresses slowly, destroying retinal photoreceptors and often leading to complete blindness by middle age.

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But the slow pace of the disorder offers a glimmer of hope by making it possible to step in with gene therapy before too much damage has occurred. The treatment involved injecting a harmless virus carrying a properly functioning copy of the CHM gene directly into the retina’s light-sensitive cells in six patients.

Publishing their findings in the New England Journal of Medicine, the experts said it was the strongest evidence to date that the effects of gene therapy could be permanent. They hope it will pave the way for a single-treatment cure for many types of inherited blindness, including retinitis pigmentosa.

Two of the patients experienced a significant improvement in vision that was sustained for at least four years. Their vision got worse in their untreated eyes. A further three patients kept their level of vision for the four years rather than losing it, while a sixth patient, who received a lower dose, experienced a slow decline in vision in both eyes.

[Prof Robert MacLaren](https://www.theguardian.com/science/2014/jan/16/sight-partly-restored-gene-therapy-oxford-hospital), the lead investigator of the study, said: “There have recently been questions about the long term efficacy of gene therapy, but now we have unequivocal proof that the effects following a single injection of viral vector [carrying the genes] are sustained.

“Even sharpening up the little bit of central vision that these patients have can give them considerable independence. Gene therapy is a new technique in medicine that has great potential. As we learn more about genetics, we realise that correcting faulty genes even before a disease starts may be the most effective treatment.”

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 Prof Robert MacLaren, centre, conducting an operation to inject a virus into the eye to deliver billions of healthy genes as part of the university’s research. Photograph: University of Oxford/PA

Gene therapy involves replacing DNA with DNA that has been programmed in the laboratory to correct faulty genes. MacLaren said: “In this case, success in getting a treatment effect that lasts at least several years was achieved because the viral DNA had an optimal design and the viral vector was delivered into the correct place, using advanced surgical techniques. In brief, this is the breakthrough we have all been waiting for.”

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Dr Stephen Caddick, director of innovation at the Wellcome Trust, which helped fund the study, said: “To permanently restore sight to people with inherited blindness would be a remarkable medical achievement. This is the first time we’ve seen what appears to be a permanent change in vision after just one round of treatment. It’s a real step forward towards an era where gene therapy is part of routine care for these patients.”

Jonathan Wyatt, the first patient in the world to be treated with the gene therapy, now has double the level of vision in his treated left eye, which has been maintained for four years so far. The 68-year-old has suffered vision problems since the age of 20. Wyatt could read 23 letters in eye chart tests before the operation but after three-and-a-half years he could read 44.

He said he felt lucky, adding: “The left eye is much improved to such an extent that I use it mostly to get about these days. It has substantially improved, it is fantastic.

“It has made me more independent, I think I would be more dependent. I think I would feel more cautious about train journeys on my own. Without it I think I would be tapping with a white stick, I think I would have remained cheerful but I would be at home more.”

Joe Pepper, 24, said: “I sat down and began the reading chart test on my treated right eye and I read the first two lines and for the first time in my memory I read on and on. I will remember that day for the rest of my life. I could see more than before the operation. I could read four lines beyond where I was earlier. I laughed and shed a tear. It was special.

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“Six months on from the surgery the results have been more than I ever imagined. My vision now has a new found clarity and I am no longer putting stress on my vision when reading or looking into the distance. Instead of looking into the distance and seeing outlines of people or trees I am seeing their defined features. At night I now have a new-found confidence in dimly lit areas that means I can feel independent and safe after dark.”

The advance was welcomed by [RP Fighting Blindness](http://www.rpfightingblindness.org.uk/), a UK charity that funds pioneering medical research and provides a range of support services to people with inherited retinal dystrophies and other related conditions.

Chief executive, Tina Houlihan, said: “This is fabulous news. The advances made in genetic therapies are extremely encouraging for people living with inherited eye conditions. We look forward to even greater steps forward being made in the future as treatments are refined ever further. Our patient community is excited so many different strands of research are proving great potential and such positive results are being reported.”