**Gene therapy reverses sight loss and is long-lasting**

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* 28 April 2016

**A genetic therapy has improved the vision of patients who would otherwise have gone blind.**

A clinical study by British scientists has shown that the improvement is long-lasting and so the therapy is suitable to be offered as a treatment.

The researchers will apply for approval to begin trials to treat more common forms of blindness next year.

The therapy involve injecting working copy of the gene into the back of the eyes to help cells regenerate.

The results of the therapy, published in the [**New England Journal of Medicine**](http://www.nejm.org/), have been tried out on 14 patients in the UK and 18 in the US, Canada and Germany over the past four and a half years.

A team at Oxford University is treating a rare disorder called **[choroideremia](http://curechm.org/)**. The disorder affects young men whose light-detecting cells in the backs of their eyes are dying because they have inherited a faulty gene.

Until now, there has been no treatment and they gradually become blind.

The researchers found that not only does the treatment halt the disease, it revives some of the dying cells and improves the patient's vision, in some cases markedly.

A 24-year-old history teacher Joe Pepper, who works at St John's school in Leatherhead in Surrey, is the latest patient to have been treated.

Joe played for Hayes Cricket Club in Kent as a boy, but was forced to give it up at 16 when his vision deteriorated.

But he could see well enough to continue his love of the game as a coach at his school.

Without the gene therapy, he would have had to give that up too within a few years, as he gradually became blind. The prospect terrified him.

"When I was 18, my vision was in a very bad place," he said. "[My sight] was degenerating at quite a speed."

He told me: "I was scared of what would have happened; I was scared of not being able to see or live the life I had.

"And now to have the belief that that's not going to happen is a weight off your shoulders."

Joe is popular with the pupils. He is jovial and laughs a lot. He teaches sport and history with a passion and enthusiasm that are infectious.

But his positive personality belies the fact that the past few years have been challenging for him as his sight worsened.

"It was really quite upsetting, not only for me, but for my family.

"I was never blind but every year there would be something new to compensate for, so my life was never steady and it was the constant changes that affected me the most, particularly when I was 18. Not being able to do what my peers were doing I found quite hard for a while."

Joe had his operation in October and began to notice an improvement soon after.

"After the operation I was looking into our garden and I could see more but I wasn't sure.

"I didn't tell my mum and dad. I didn't want to let anyone get excited until we had done a simple vision test in a week's time."

The test involved reading a sight chart of letters. Each line on the chart had progressively smaller letters.

He read line after line, going four lines beyond where he had ever read before. The medical staff were astounded.

"Everyone in the room just looked at me and I looked at them," he said reliving the moment.

"Anna, the research nurse, was taking me back out of the waiting room and she just looked at me and said it was 'fantastic' - both of us just had tears in our eyes."

Joe met his father in the waiting area. He was alarmed at the sight of his son in tears.

"He thought that it was [bad news] and he just looked at me and we both sat there in each others' arms just realising that actually it had..." [Joe could not finish his sentence].

It was only when he was giving me his account of the joy and relief he felt with his father that the full emotional impact of the moment hit him for the first time.

After a brief pause he continued: "I cannot explain how terrifying and upsetting it was when I was younger," he continued.

"And now to know that there is so much opportunity, there is so much that I can actually do and do the things that I have actually wanted to and continue to do the things I really enjoy."

## Early promise

There are others with similar stories to Joe's.

Wayne Thompson, a 46-year-old IT worker from Staffordshire was delighted to be able to see stars in the night sky for the first time following his operation. Jonathan Wyatt, a 68-year-old former lawyer, says "it has opened a new chapter of my life for me when I thought that the book was about to close".

As the researchers have gained confidence in their treatment, they have tried it out on progressively younger patients who still have reasonably good vision.

Joe is the youngest and the most successful so far. This suggests that the gene therapy may be most effective on younger patients before the disease becomes irreversible.

The study also indicates that the treatment is long-lasting. The first patient received the therapy four and a half years ago and his treated eye has shown no drop-off in vision. Indeed, there are still slight improvements in his vision in that eye.

## 'Corrects defects'

This is the first indication that the treatment is viable and could be widely used on patients, according to the eye surgeon leading the trial, Prof Robert MacLaren of Oxford University.

"The concept of gene therapy is that it corrects gene defects. Ideally, we should only have to do that once, because once the DNA is corrected and inserted into the correct cell, that cell should be able to continue its function as normal," he told BBC News.

"We seem to have achieved this concept of one single treatment that does not need to be repeated which is unlike traditional medicines."

Prof MacLaren says that if the next phase of larger trials goes as he anticipates, a gene therapy for choroideremia will be licensed in three years.

He has also begun to develop gene therapy trials to treat more common forms of blindness, such as retinitis pigmentosa and macular degeneration. These could begin as early as next year.

Treating these disorders will be more challenging. Choroideremia is caused by a defect in a single gene, whereas the more common forms of blindness involve several.

But Prof MacLaren believes that the choroideremia trials have laid the ground for the next phase of studies and, crucially, shown that gene therapy for blindness is safe and works.

"When I started my career as an eye surgeon when we had these patients that had inherited diseases, not only did we tell them nothing could be done but we would actually discharge them from the clinics.

"We are now calling them back in to test them, to look at them in great detail because potential treatments are available. To treat a disease at the genetic level is surely the most efficient way of treating a disease, to prevent it from happening in the first place.

"We would like to develop treatments for more common forms of blindness and this may be available in the next five to 10 years," he told BBC News.

The Research is funded by the [**Health Innovation Challenge Fund**](http://www.wellcome.ac.uk/Funding/Innovations/Awards/Health-Innovation-Challenge-Fund/index.htm) which is a partnership between the Wellcome Trust and the Department of Health.