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Gene therapy success 'reverses' blindness
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Experimental gene therapy trials have improved the vision of four people who suffer from hereditary blindness.

The preliminary results of two independent studies suggest that "repair" genes delivered to the eye might one day cure Leber Congenital Amaurosis (LCA), a rare disease that strikes about 1 in 80,000 people in the UK, and 2,000 Americans in total.

Equally important, say researchers, the treatments proved safe in the six patients who received the genes – delivered by a disabled virus – via eye surgery.

"This is really an exciting result for gene therapy as a field," says Katherine High, of the University of Pennsylvania Medical School in Philadelphia, who was part of an international team that presented the findings at a conference yesterday.

Another team led by Robin Ali, of University College London, presented similar results.

Dog success
In High's trial, the vision of all three patients improved noticeably, while one of Ali's patients saw well enough to navigate an obstacle course in dim light – a task that had been a struggle before the treatment.

While extremely rare, LCA is a debilitating disease that strikes patients at birth. As their retinal eye cells die off, most patients become completely blind by their 30s.

Mutations in at least six genes cause LCA, but both teams treated patients with a mutated version of a gene called RPE65, which is responsible for about 6% of cases.

In 2001, scientists reversed the blindness of dogs born with the same mutated gene. A harmless virus called adeno-associated virus injected a working copy of RPE65 to the animal's retinal cells, kicking them back into action.

Speedy improvement
After proving the treatment safe in other animals, including primates, each team gave the modified virus to three LCA patients in their late teens and early 20s.

"They were not completely blind, but they were severely visually impaired," says Jean Bennett, a colleague of High's at the University of Pennsylvania. For instance, patients could see a hand waving in front of their face, but most had trouble reading any letters on an eye chart.
After receiving surgery to inject the virus into one eye – the weakest – all three of Bennet and High's patients noticed quick improvement.

They saw better in dim light, and two patients could now read the first three lines on an eye chart. One patient, who fumbled through an obstacle course before the surgery, had few problems navigating after treatment.

Youngest benefit
So far only the youngest of the three subjects in Ali's trial, an 18-year old man named Steven Horwath, has had improved vision after surgery. "Before the operation, I used to rush home from college when it started to get dark because I was worried about getting around," he says. "Now I can take my time and stay later

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at college if I need to, for band rehearsals and things like that," he says.

Less subjective tests that gauge the eye's response to a tiny flash of light also indicated that patients' treated eyes had improved.

Both teams plan to test the therapy on younger patients who might regain even more of their sight. "I think the effect will be most dramatic in younger individuals, when the retina has not degenerated so extensively," Bennett says.

Turning point

The treatment also seems safe. Patients in both studies showed little immune response to the virus, a problem in some previous gene therapy trials. And the virus seemed not to stray from the eye region.

One patient in Bennett and High's study, a 26-year old male, developed a microscopic hole in his retina after surgery. The treatable complication didn't worsen his vision, but it could be a problem for younger LCA patients with better sight, says Joan Miller, a retina specialist at Harvard Medical School in Boston, not involved in either study.

But the success of both studies should buoy gene therapy's troubled past. "I think it could be a real turning point," Miller says.

Last year, a patient enrolled in a gene therapy trial to treat her rheumatoid arthritis, died – although regulators say the therapy probably didn't cause the patient's death. And in 1999, 18-year-old Jesse Gelsinger died after receiving gene therapy to cure a rare metabolic disease.

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