Gene therapy improves eyesight in people born with an incurable form of blindness

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A new gene therapy has restored some sight in people born with an inherited, progressive form of blindness. The technique replaces a defective gene in the eye with a normal working copy of the gene using a single injection.

The findings of this phase 1 trial, published in *The Lancet*, support the further development of this state-of-the-art treatment for other more common genetic causes of blindness including degenerative diseases of old age, such as macular degeneration, and inherited defects like retinitis pigmentosa.

Choroideremia is caused by a mutation in the CHM gene on the X chromosome and affects an estimated 1 in every 50 000 people. The condition causes progressive loss of vision due to degeneration of the choroid, retinal pigment epithelium, and retina. There is no treatment for choroideremia and eventually the photoreceptor cells—the rods and cones in the retina that respond to light by sending signals to the visual processing areas of the brain—also degenerate leading to complete blindness by middle age.

"The cellular degeneration in this disease is fairly slow, providing a reasonably large window of time in which to intervene before the onset of visual loss", explains Professor Robert E MacLaren from the University of Oxford in the UK who led the research.

MacLaren and colleagues assessed the effect of gene therapy on retinal and visual function in six patients aged 35–63 years with different stages of choroideremia. They injected patients' retinas with a vector—a genetically engineered adeno-associated virus (AAV)—to deliver a corrective copy of the gene into the appropriate part of the eye to halt photoreceptor cell death.

The treatment caused no harm and resulted in improvements in subjective measurements of vision. Six months after delivery of the gene, all patients recovered their visual acuity from before the procedure, and two patients showed substantial improvements in how well they could see, with one reading over three additional lines on an eye chart. Importantly, the patients developed increased sensitivity to light, compared to a loss in sensitivity in the untreated eyes.

According to Professor MacLaren, "This is first time that gene therapy has been used to treat patients with normal visual acuity before the onset of clinically significant retinal thinning. Our findings hold great promise for gene therapy to prevent loss of sight in other retinal diseases such as age-related macular degeneration."

Writing in a linked Comment, Hendrik Scholl from John Hopkins University, Baltimore, USA, and José Sahel from the Institut de la Vision, Paris, France, say, "The ultimate goal of gene therapy in choroideremia would be to save the central retina from further degeneration. The short follow-up of the new study prevents any conclusion about preventing degeneration in the long term; indeed, results from the morphological assessment suggested that degeneration is rather continuous…It remains to be determined if gene therapy targeting REP1 will have an effect on the progression of photoreceptor degeneration. Even if the effect turns out to be only slight, this might have important positive implications because there are additional therapeutic avenues targeting photoreceptors that could help to save or restore visual function."

More information: Paper: www.thelancet.com/journals/lan … (13)62117-0/abstract

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