

Gene therapy for choroideremia using an adeno-associated viral vector encoding Rab escort protein 1: the REGENERATE open-label trial

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Plain language summary

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Plain language summary

Choroideremia is a rare eye disease caused by a defective gene that prevents the cells in the retina, the light-sensitive layer at the back of the eye, from functioning normally. The disease usually affects males, although females can also develop the disease. In childhood, choroideremia patients initially experience 'night blindness', or difficulty in seeing in low light. As the disease progresses, there is a gradual loss in the peripheral vision, eventually resulting in 'tunnel vision' and finally in complete blindness by late adulthood.

The aim of this study was to investigate if gene therapy was able to prevent further vision loss in patients suffering from choroideremia. The gene therapy for choroideremia uses a modified non-pathogenic virus, called a viral vector, to carry healthy copies of the affected gene into the retinal cells and thereby help the cells to function normally. Thirty male participants from two NHS eye hospitals took part in the study. Each participant received gene therapy in one eye, with the other eye left untreated for purposes of comparison. The condition and function of the treated and untreated (control) eyes were then monitored at regular time points over a 24-month period.

A meaningful difference in the comparative rate of vision loss in the treated and control eyes was not observed during the 24-month assessment period of this study. However, it should be noted that choroideremia is a very slow degeneration and vision loss in the control eyes did not decline significantly during this period. Three participants experienced worse vision in their treated eyes, including one case of sight loss caused by severe inflammation that did not respond to medication. Additional data are being collected in a follow-on observational study to track the long-term progress of retinal degeneration in the treated eyes and control eyes of the participants.

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