Long-term efficacy of AAV5-based gene therapy to treat day blind sheep with achromatopsia

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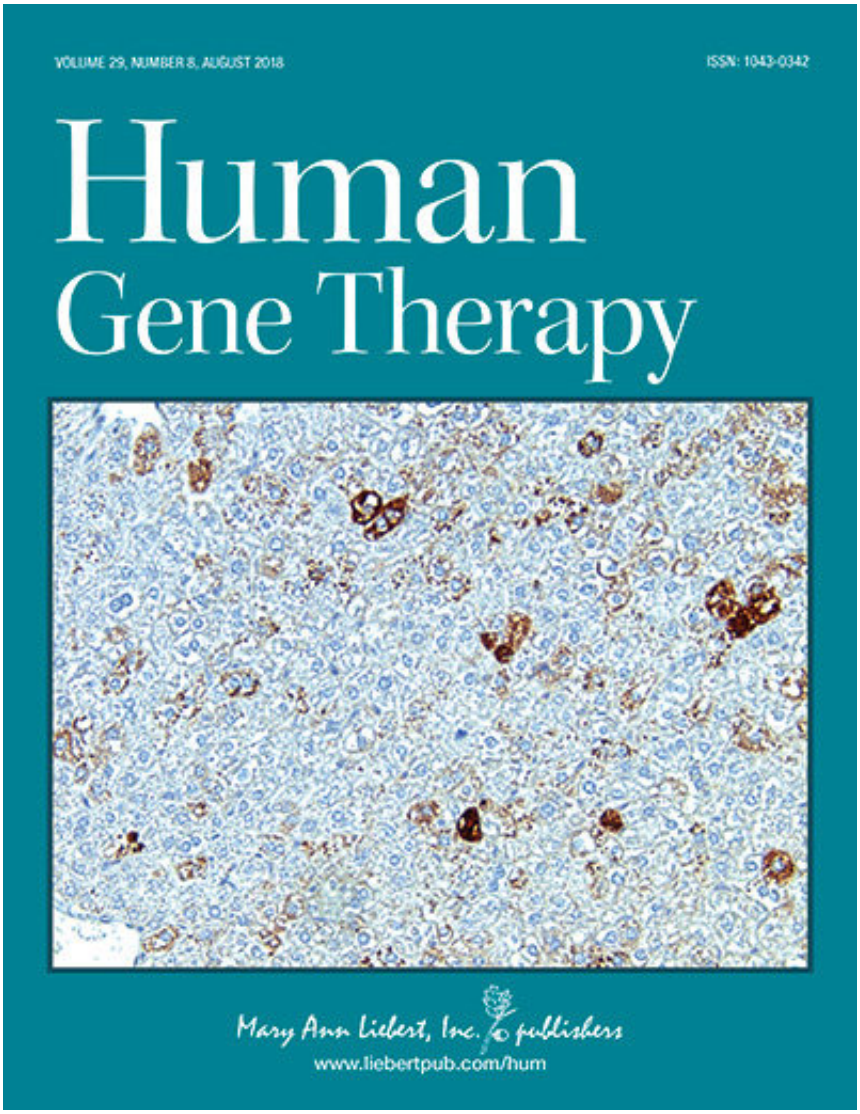

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A study of a large animal model of achromatopsia caused by a mutation in the CNGA3 gene that was treated with a single injection of CNGA3 gene therapy delivered using an AAV5 vector revealed findings reported long-term follow-up findings that show promise for the efficacy and safety of this therapeutic approach. The results demonstrated significant improvement in cone function and no abnormalities in the treated eyes for up to six years following delivery of the CNGA3 transgene, as reported in an article published in *Human Gene Therapy*.

"Six Years and Counting: Restoration of Photopic Retinal Function and Visual Behavior Following Gene Augmentation Therapy in a Sheep Model of CNGA3 Achromatopsia" describes the recovery of cone photoreceptor function, as determined by photopic behav-ioral maze tests and electroretinographic (ERG) examinations. The treated sheep had pas-sage times on the photopic maze test and numbers of collisions that were significantly lower than pre-treatment values at all follow-up exams. ERG testing showed significant improvement throughout the follow-up period.

Ron Ofri, Koret School of Veterinary Medicine, The Hebrew University of Jerusalem (Rehovot, Israel) and Edward Averbukh, Hadassah-Hebrew University Medical Center (Jerusalem, Israel) led a team of researchers from The Hebrew University of Jerusalem, Hadassah-Hebrew University Medical Center, Agricultural Research Organization (Beit Dagan, Israel), and University of Florida (Gainesville) as coauthors of the article.

"Confirmation of the concept of 'once-in-a-lifetime' gene therapy for genetic blindness depends on patient, careful studies such as these," says Editor-in-Chief Terence R. Flotte, MD, Celia and Isaac Haidak Professor of Medical Education and Dean, Provost, and Executive Deputy Chancellor, University of Massachusetts Medical School,

Worces-ter, MA. "Human gene therapy investigators in the field of retinal diseases should find these results very encouraging, as they move forward in expanding clinical applications of the platform of rAAV gene therapy."

More information: Ron Ofri et al, Six Years and Counting: Restoration of Photopic Retinal Function and Visual Behavior Following Gene Augmentation Therapy in a Sheep Model of CNGA3 Achromatopsia, *Human Gene Therapy* (2018). DOI: 10.1089/hum.2018.076

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