Research to Prevent Blindness

NEWS

FALL 2018

Gene Therapy for Retinal Disease

In March, a 13-year-old boy, Jack Hogan, was the first patient to receive an FDA-approved gene therapy for inherited blindness called Luxturna. The treatment, administered by RPB-supported researchers and physicians at Massachusetts Eye and Ear, is designed to improve visual function in children and adults with inherited retinal disease caused by mutations in the gene RPE65. The lead surgeon is an RPB Career Development Awardee, Jason Comander, MD, PhD, Associate Director of the Inherited Retinal Disorders Service at Mass. Eye and Ear.

According to Mass. Eye and Ear, Luxturna involves injecting a modified virus into a patient's eyes to correct a deficiency caused by mutations in the RPE65 gene. These mutations prevent the production or function of a protein needed for proper functioning of the photoreceptors, the light-sensitive cells in the back of the eye that initiate vision.

Since receiving his treatment, Hogan has experienced a significant impact on his quality of life. He's enjoying better vision in low light, such as when playing basketball outside in the evening with friends or going to the movies, and is reading 40 percent smaller print.

"It is truly amazing to see large improvements in Jack's vision, which would have been impossible without this treatment," said Dr. Comander. "It is going to make a big difference in his life. These results are representative of just how big a moment this is for gene therapy; it is helping our patients and it is here to stay."

Photo credit: Massachusetts Eye and Ear

Dr. Jason Comander (left) of Massachusetts Eye and Ear with his patient Jack Hogan, who received groundbreaking gene therapy treatment for an inherited retinal disease.

Did You Know?

Photo credit: AMR Image

Risk factors for glaucoma include:

- **having a family history of the disease,**
- **being over age 35,**
- **having diabetes or hypothyroidism,**
- **being nearsighted, and**
- **being of African-American or Hispanic heritage.**

Learn more about glaucoma risk factors and what you can do to mitigate your risk with RPB's Glaucoma Fact Sheet, available online at http://bit.ly/glaucoma18.

Ocular Cancer Screening

An RPB-supported clinician, Patricia Chévez-Barrios, MD, at the Baylor College of Medicine in Texas, was a key member of a consensus group that developed new surveillance guidelines for children at risk for retinoblastoma, an eye cancer most common in early childhood. A patient "at risk" was defined as a person with a family history of the cancer in a parent, sibling or first- or second-degree relative.

The consensus group developed five recommendations:

1. Dedicated ophthalmic screening is recommended for all children at risk for retinoblastoma above the population risk.
2. Frequency of examinations should be adjusted on the basis of expected risk for RB1 mutation.
3. Genetic counseling and testing can clarify the risk for retinoblastoma in children with a family history of the disease.
4. Examination schedules are stratified on the basis of high-, intermediate- and low-risk children.
5. Children at high risk for retinoblastoma require more frequent screening, ideally as examinations under anesthesia.

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Novel AMD Therapy

RPB-supported researchers and physicians at the University of Southern California (USC) Roski Eye Institute have collaborated with other California institutions to show that a stem cell-based retinal implant is feasible for use in people with advanced dry age-related macular degeneration (AMD).

The treatment, which consists of a layer of stem cell-derived retinal pigment epithelium cells on an ultrathin supportive structure, was implanted in the retina of four patients by a USC Roski Eye Institute surgeon. The patients were followed for up to one year to assess its safety and the treatment was well-tolerated. There was also evidence that the implant integrated with the patients' retinal tissue, which is essential for the treatment to work. Preliminary assessments of the therapy's efficacy showed that one patient had improvements in visual acuity, while two showed gains in visual function.

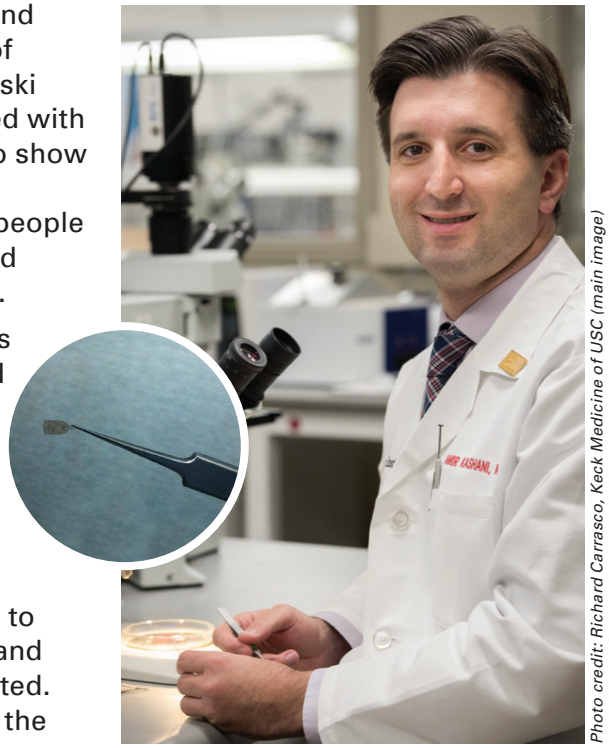

Photo credit: Richard Carrasco, Keck Medicine of USC (main image) and Britney O. Pennington, PhD (insert image)

Amir Kashani, MD, PhD, lead author and surgeon for the study, in his lab. The inset image shows the implant used in the study.

AMD's Impact

AMD not only affects the ability to see, it can affect the ability to perceive as well. RPB-supported researchers at The Johns Hopkins Wilmer Eye Institute in Maryland conducted tests of reading speed and reading comprehension in people with and without AMD. The study showed that people with AMD demonstrate substantially lower reading comprehension when reading at speeds similar to people without the condition. The findings underscore the need for innovative AMD treatments such as the one outlined above.

A GIFT TO RPB CAN SAVE SIGHT

Research to Prevent Blindness, Inc. (RPB) is the only public foundation supporting research aimed at treating, preventing or curing all diseases that damage and destroy vision. Your support is critical to the success of our efforts. Contributions totaling up to one million dollars within a calendar year are matched, effectively doubling your gift. ALL GIFTS AND BEQUESTS ARE TAX DEDUCTIBLE.

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