

Usher Syndrome Gene Therapy Clinical Trial Underway

The first-ever gene therapy for Usher syndrome is in a Phase I/IIa clinical trial at the Foundation-funded Casey Eye Institute, Oregon Health & Science University (OHSU), in Portland, and the Centre Hospitalier National d'Ophtalmologie des Quinze-Vingts in Paris, France. Known as UshStat®, treatment is being developed by the pharmaceutical company Sanofi. It's designed to halt vision loss in people affected with Usher syndrome type 1B, which is caused by defects in the MYO7A gene. Based on results in lab studies, researchers believe a single UshStat treatment may last several years, perhaps a lifetime. The Foundation Fighting Blindness is funding the Paris site of the clinical trial and funded lab research that made the human study possible.

Dietary Supplements Slow Vision Loss

Researchers funded by FFB report that a regimen consisting of vitamin A palmitate supplementation,

consumption of oily fish high in the omega-3 fatty acid DHA, and lutein supplementation may slow loss of vision in people with retinitis pigmentosa (RP) and Usher syndrome types 2 and 3. Good sources of DHA include: salmon, tuna, mackerel, sardines, and herring. Download the [Update on Research for RP Treatment: Vitamin A Palmitate, Omega-3-Rich Fish, and Lutein](#) from the Foundation's web site for more information.

Gene Therapy Revives Cones Long After They Stop Working

A Foundation-funded research collaboration from the Institut de la Vision in Paris and the Friedrich Miescher Institute in Basel, Switzerland, is developing a gene therapy that revives degenerating cones, enabling them to regain their ability to respond to light and provide vision. The treatment also improves the health of cones and extends their lifespan significantly. Cones are the retinal cells that allow people to see color and fine

detail, enabling them to drive, read and see the faces of loved ones. A key benefit of the approach is that it may help people affected by a range of conditions, including many forms of retinitis pigmentosa and Usher syndrome, because it works independently of the underlying disease-causing genetic defect. The collaboration's goal is to move the gene therapy into a clinical trial within three years.

Gene Therapy Successful in Usher Syndrome Type 2D Model

Researchers from the University of Utah used gene therapy to correct the disease-causing genetic defect in the retinas of mice with Usher syndrome type 2D (USH 2D). Led by Dr. Jun Yang, the Foundation-funded research team developed a manmade virus to deliver copies of normal USH 2D genes to the photoreceptors of the affected mice. Tests revealed that copies of the therapeutic USH 2D gene reached photoreceptors and restored function of the cilia. While more laboratory work needs to be done, the advancement is an important step toward moving the treatment into a clinical trial.

Emerging Treatment for Retinal Diseases Gets the Message Right

In about 12 percent of all retinal degenerative diseases, the translation of genetic messages (mRNA) for the production of critical proteins stops prematurely, leading to the production of nonfunctional proteins, and resulting in vision loss. In simple terms, it's as if someone stops reading a sentence halfway through, and the resulting message doesn't make sense. These translational errors are due to what is known as premature termination codons or PTCs. In a Foundation-funded study at the Johannes Gutenberg University in Mainz, Germany, Uwe Wolfrum, Ph.D., and his team are evaluating a drug that can "read through" PTCs in retinal cell cultures and mouse models of Usher syndrome type 1C. The drug enables the cell to read the complete message and make the right protein.

Foundation Commits \$2 Million to Development of a Cross-Cutting Drug Treatment

The Foundation Fighting Blindness has invested more than \$2 million in MitoChem Therapeutics, a start-up company which, thanks to prior Foundation support, has identified

compounds that appear to boost mitochondrial function and show potential for significantly slowing vision loss caused by a variety of retinal degenerations. Mitochondria are the power supplies for all cells. The goal is to determine which compound will work best in people and move it into a clinical trial.

Company Launches Stem Cell Clinical Trial

ReNeuron, a stem-cell therapy development company in the United Kingdom, has received authorization from the U.S. Food and Drug Administration to launch a Phase I/II clinical trial of its stem cell therapy. The company is partnering with the Schepens Eye Research Institute, Massachusetts Eye and Ear Infirmary, to develop the treatment. The emerging treatment involves the transplantation of retinal progenitor cells — cells which haven't completely developed into photoreceptors. The emerging

therapy may benefit people with retinitis pigmentosa, Usher syndrome, and other retinal diseases. The Foundation funded decades of critical lab research to make this emerging therapy possible.

The Foundation publishes frequent updates on the latest advancements in research and clinical trials for Usher syndrome and similar diseases.

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