FOUNDATION FIGHTING BLINDNESS

RETINITIS PIGMENTOSA
RESEARCH ADVANCES

Landmark gene therapy restores vision in children and young adults

More than 40 children and young adults who were virtually blind have had some vision restored thanks to an innovative gene therapy to cure a severe form of retinitis pigmentosa known as Leber congenital amaurosis (LCA). The individuals are participating in FFB-funded Phase I clinical trials of the treatment at The Children's Hospital of Philadelphia (CHOP) and Moorfields Eye Hospital in London, as well as a study at the Universities of Pennsylvania and Florida. One nine-year-old boy has put away his white cane and can now see the blackboard at school. A young woman was able to see fireflies for the first time after receiving the treatment. Participants in the study at CHOP are now having their second eyes treated. Success in these studies is paving the way for development of gene therapy to treat a variety of retinal degenerative diseases. The Foundation funded much of the preclinical research that made these clinical trials possible.

FDA-approved drug moving into clinical trial for dominant retinitis pigmentosa

Valproic acid, a drug which has shown promise for preserving vision in people affected by autosomal dominant forms of retinitis pigmentosa (adRP), is in a Foundation-funded multicenter Phase II clinical trial. The drug is already FDA approved for seizure disorders. The three-year, 90-participant clinical study of valproic acid is being conducted under the auspices of the National Eye Evaluation Research Network. The Foundation established the network to launch clinical trials of promising treatments and cures for retinal degenerative diseases. Valproic acid is the first treatment to be evaluated in the network.

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). Good sources of DHA include: salmon, tuna, mackerel, sardines, and herring. For more information (including dosing and contraindications) on the regimen, see the following link: http://www.blindness.org/rp-nutrition/index.asp.

QLT Conducting Clinical Trial of RP and LCA Treatment

The biopharmaceutical company QLT is conducting an international clinical trial for its synthetic retinoid treatment for people with Leber congenital amaurosis (LCA) and retinitis pigmentosa (RP) caused by variations in the RPE65 or LRAT genes. Individuals with these genetic variations do not produce a retinoid critical for vision; QLT's treatment serves as a replacement for that missing retinoid. QLT announced impressive results for their Phase IB clinical trial for people with LCA and RP. Patients responded well to the seven-day treatment, reporting some gains in acuity and/or size of their visual field. The investigators were encouraged that the effect of the week-long oral treatment persisted for several months.

Recessive RP Gene Therapy Clinical Trial Underway in Saudi Arabia

An international research team has launched a Phase I clinical trial of gene therapy to treat people with autosomal recessive retinitis pigmentosa caused by variations in the MERTK gene. The trial will be taking place in Saudi Arabia. The investigative team includes FFB-funded scientists Khang Zhang, M.D., Ph.D., of the University of California, San Diego, and William Hauswirth, Ph.D., of the University of Florida. Investigators will be using the same type of gene delivery mechanism — an adeno-associated virus or AAV — currently being utilized in gene therapy clinical trials underway for Leber congenital amaurosis.

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, 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 for Autosomal Dominant Retinitis Pigmentosa

Researchers from the University of Florida are developing a gene therapy for people affected by autosomal dominant retinitis pigmentosa (adRP) caused by variations in the Rhodopsin

gene. A challenge in creating a gene therapy for adRP is that, in addition to delivery of a healthy gene, the bad gene needs to be turned off. In some cases of adRP, the researchers demonstrated that through a one-step process they can simply override the bad gene with a healthy gene. In other cases, they will have to use a two-step process — "knocking down" the bad gene and delivering a healthy gene. If successful, this work positions them well to obtain FDA authorization to launch a clinical trial.

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

The Foundation Fighting Blindness is giving $2 million to MitoChem Therapeutics, a start-up company which, thanks to prior Foundation support, has identified three compounds that appear to boost mitochondrial function and show potential for slowing vision loss caused by a variety of retinal degenerations. The goal is to determine which one will work best in people and move it into a clinical trial.

A key to survival for any organism, plant or animal, is energy. And, in humans, every cell gets its energy from a tiny, organ-like structure called a mitochondrion. It operates like a power plant, providing the energy needed to stay alive and functioning. Among their many functions, mitochondria combine sugar and oxygen, which serve as the cells' supply of fuel.

One consequence of most retinal degenerations, including retinitis pigmentosa and macular degeneration, is that mitochondria operate at reduced capacity, because of disease-related stress. Ultimately, photoreceptors, the cells in the retina that provide vision, are lost.

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

www.FightBlindness.org