

**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) caused by mutations in the RPE65 gene. The individuals are participating in clinical trials of the treatment at The Children's Hospital of Philadelphia (CHOP), Moorfields Eye Hospital in London, the Universities of Pennsylvania and Florida, and other centers around the world. 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. The CHOP clinical trial has now advanced to Phase III and expanded to the University of Iowa. CHOP has also spun off a

company, Spark Therapeutics, to commercialize the treatment and gene therapies for other ocular diseases. The Foundation funded much of the preclinical research that made these LCA gene-therapy clinical trials possible.

**FDA-Approved Drug in Clinical Trial for Dominant Retinitis Pigmentosa**

Valproic acid, a drug which has shown potential 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 90-participant clinical study of valproic acid is being conducted under the auspices of the Foundation's Clinical Research Institute. The Foundation established the institute to launch clinical trials of promising treatments and cures for retinal degenerative diseases.

Valproic acid is the first treatment to be evaluated by the institute.

### **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. 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.

### **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. Though the primary goal of this early-stage trial is to establish safety, some participants have reported modest vision improvements.

### **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 they can simply override the bad gene with a healthy gene through a one-step process. In other cases, they will have to use a two-step process — “knocking down” the bad gene and delivering a healthy gene. This work is positioning 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 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 Plans RP Stem Cell Clinical Trial**

ReNeuron, a stem-cell therapy development company in the United Kingdom, has been granted an orphan designation by the

U.S. Food and Drug Administration (FDA) and the European Commission for its emerging retinitis pigmentosa treatment known as ReN003.

The designation bolsters ReNeuron's plan to launch a Phase I/II clinical trial for ReN003 in 2014. 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, which are more mature than embryonic stem cells, but haven't completely developed into photoreceptors, the cells in the retina that make vision possible. The Foundation funded critical lab research that made this emerging therapy possible.

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

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