Odylia Therapeutics is a nonprofit working to accelerate thedevelopment of therapies for people with rare diseases by changing the way treatments are brought from the lab to patients regardless of prevalence or commercial interest. By shifting the paradigm to focus on proven science and removing roadblocks to therapeutic development, Odylia can change the lives of those with rare diseases.

Leber Congenital Amaurosis (LCA) and the RPGRIP1 Mutation

Leber congenital amaurosis (LCA) is a rare type of inherited eye disorder that primarily affects the retina and leads to vision loss at birth or in early childhood.

- Accounts for 20% of blindness in school-age children
- Vision-loss onset often seen at birth
- Symptoms can include profound vision loss, involuntary eye movements (nystagmus), crossed eyes (strabismus), and sensitivity to light (photophobia)

At least 27 different genes are associated with LCA. Mutations in both copies of the RPGRIP1 gene cause a spectrum of vision-related deficits most commonly diagnosed as LCA type 6 (LCA6). LCA6 patients are often completely blind from early childhood and have a rapid decline in function of the photoreceptor cells of the retina. The RPGRIP1 protein is needed for the normal function of the photoreceptors (rods and cones) of the retina, and loss of this protein is what causes vision loss in patients. LCA6 is particularly devastating due to the rapid onset and progression of vision loss.

Patients with LCA6 describe vision loss (photo on the right):

Gene Therapy for LCA6

Odylia is developing a gene therapy to treat vision loss caused by RPGRIP1 mutations, for which there is currently no treatment. This gene therapy uses the novel Anc80 vector and builds upon proof-of-concept data generated at Massachusetts Eye and Ear in the labs of Eric Pierce and Luk Vandenberghe.

Together we can prevent vision loss

To date, Odylia's studies have demonstrated that the RPGRIP1 protein localized to the appropriate part of the photoreceptor/retinal structure and there was a clear dose-dependent increase in expression of RPGRIP1 in the mouse model. Additionally, the gene therapy demonstrated effectiveness through protection against retinal degeneration and preservation of outer nuclear layer and inner nuclear layer thickness as well as improved ERG.

Getting this treatment to clinical trials will cost approximately $4.3 million. Donations are sought to support this program. Treating rare diseases, making a meaningful impact on the lives of patients, is going to take many of us. Each one, doing their part to reach the goal.

For more information about how you can help, contact us at hgreene@odylia.org.