Letter from the President

Dear PRRF Community,

Our organization has been very successful in its first years. One of the tasks as we grow is to identify the next challenges and goals. As I often tell physicians-in-training - and my kids - if you are hardworking and passionate about what you do, there is a very good chance you will achieve the goals you set for yourself. The real challenge, therefore, is in picking the goals.

Our main goal in 2020 from a mission perspective is to “build community and build capacity” - get the PRRF message out to families and the broader community. We will also be working to bolster our social media initiative and undertake production of an informational video as two steps towards that community building goal. We are working also to develop a programmed - giving initiative for families and trusts to complement our current event-oriented fundraising initiatives.

This issue of Sightlines will catch you up on some of our activities to date and provide information which we hope you will find interesting. In our personal focus section, we are running with a theme for 2020, highlighting young adults in our community. Dr. Drenser provides a brief update on gene therapy. On the fundraising front, we are off to another great year – as Kelli Matthews details in “Celebrating Our Generous Donors” below.

Heartfelt thanks to the dedicated and talented members of our PRRF community who share the passion for working on behalf of “our kids”.

Antonio Capone, Jr. MD
President, The Pediatric Retinal Research Foundation (PRRF)
Campaign Update

Celebrating Our Generous Donors

We are pleased to report that our 2019 year-end capital campaign was a huge success! We raised over $50,000 from our PRRF community and William Beaumont Hospital in Royal Oak, Michigan has committed, through the Children’s Miracle Network, the remainder needed to purchase a hand-held Optical Coherence Tomography or “OCT.” Historically, to diagnose retinal diseases, physicians use an upright OCT that required patients to sit upright, leaning forward with their chin resting on a strap, and eyes fixated on a visual target – not practical for infants and children. This handheld OCT will enable physicians to use advanced technology to evaluate children with pediatric diseases while they are laying down and asleep under anesthesia. We are grateful to all our PRRF donors for their generosity which enabled us to put into practice a game changer in the management of pediatric retinal diseases. Simply put – thank you!

~Written by Kelli Matthews

On The Research Front

Update on Gene Therapy

The FDA approval of Luxterna, the first gene therapy approved for the retina, created a wave of excitement and investment dollars in therapies involving gene editing.

Traditionally, gene therapy is really gene replacement, where a virus acts as a slave “Trojan horse” to deliver a normal (non-mutated) gene into the host cell’s chromosome. That is exactly what Luxterna does. It uses an adeno-associated virus (AAV) to introduce a healthy RPE65 gene into the retinal cells for the treatment of Leber’s Congenital Amaurosis (LCA), which leads to complete blindness in early life. This technology does not eliminate the damaged gene but works by introducing one healthy copy, which is enough to overcome the disease. That approach is being used to target similar gene therapies for X-linked Congenital Retinoschisis and Choroideremia. All of these therapies use a form of engineered AAV to introduce the new gene.

The technology is elegant in its simplicity. Essentially, the “guts” of the virus are removed and replaced with the gene of choice. The “shell” remains and allows the new, recombinant virus (rAAV) to infect a cell and introduce the healthy gene into the chromosome. This technology works well for a small gene (only 5,000 nucleotides can be packaged into rAAV) and a disease where one healthy gene is enough to override the deficient gene.

So, what can be done to correct mutations that affect large genes (more than 5,000 nucleotides) or have a dominant effect (meaning that one bad copy of the gene is enough to cause disease)? That requires a type of DNA correction known as gene editing. It is still a viral-based system, most using rAAV, but instead of introducing a gene, it introduces a type of splicing that allows for the repair of the damaged DNA. Think of it as scissors cutting a ribbon and replacing a damaged part of the ribbon with a new piece. In short, it exchanges one nucleotide for another, thereby repairing the DNA and changing the damaged gene into a healthy gene. This is in clinical trials for another form of LCA that affects the CEP290 gene. The study gene therapy is EDIT-101 and is the first retinal gene therapy employing CRISPR technology.

These are exciting times for the advancement of gene therapies and, hopefully, represents the tip of the iceberg.

~Written by Kim Drenser, MD, PhD
Calvin Ventresca is an outgoing, cheerful, and forward-thinking 10th grader, making use of his talents with limited sight. Calvin was born with Congenital X-Linked Retinoschisis. He explained that he looks “normal”. No one would guess that he cannot read fine print and is legally blind in both eyes. He has gone through multiple surgeries to maintain his vision in his left eye, (he was born with a macular scar in the right), and productively utilizes all the resources at his school to continue his education.

His mother, Lilia, explained that early intervention made such a difference. They were recommended to Dr. Drenser, who has been taking care of Calvin’s eyesight. Lilia also said that support systems are critical and sometimes a parent has to search carefully to find the resources and accommodations a child needs. Calvin has an excellent teacher consultant for the visually impaired who has read tests to him, arranged for a talking iPad, and helped make schoolwork more manageable. Lilia said that the state of Michigan has some of the best programs in the country for the visually impaired.

When asked about all his operations, Calvin said, “I had to do the surgeries” to maintain his vision so he did not worry about them. At this moment, his vision is stable and he about to get his driver’s license. Calvin feels that his impairment has given him the strength of adaptability. He explained that he often cannot do things in the “normal” way that a sighted person would, and he enjoys the challenge of accomplishing what he wants to through his own ideas. He thinks it is “cool” and “fun” to look ahead to new ways of managing his life. Calvin is very open about his disability and willing to share his story. He would like others with disabilities to not think they will not succeed because of their situations but to have a belief that they will prevail.

~ Written by Kay White and photograph provided by Lilia Ventresca
Save The Date!

Dr. Patrick Droste and his team will again spearhead the **CARS FOR A CAUSE** event in SW Michigan on **July 25, 2020**. A great way to spend the day, see some incredible cars, and meet the Pediatric Retinal Research Laboratory staff for an up close and personal discussion of what's going on in the PRRF sponsored lab at Oakland University.

We will also have the second annual **HOPE FOR VISION WALK** on Sunday, **September 13, 2020** in Birmingham, MI. It’s sure to be a fun fall event as well, with a Zumba warm-up, music, food and a chance to walk through the lovely Bloomfield Village neighborhood.

Help us support families impacted by blinding pediatric retinal diseases and champion the quest for a cure. Visit [https://www.pediatricrrf.org/donate](https://www.pediatricrrf.org/donate).

To receive this newsletter electronically please send an email to info@pediatricrrf.org and write “newsletter” in the subject line.

PRRF | info@pediatricrrf.org | www.pediatricrrf.org