Letter from the President

Wow, what a year!

A year ago at this time our year-end fund-raising initiative was just underway. Then 2019 began with a bang as we exceeded our fundraising goal for the purchase of a new DNA sequencer. We also held two successful inaugural fund-raising events in 2019: the “Cars for a Cause” fund-raiser under the leadership of Board member Dr. Patrick Droste and his team, and our first “Hope for Vision Walk” this September (see the story by Traci Martin, below). Then there are those in our community who are taking a personal initiative to fund-raising in fun and innovative ways, as you will learn as you read about young Rebecca in our “On a Personal Note” spotlight below.

We’re proud to inform our supporters in the PRRF community that the Pediatric Retinal Research Laboratory (PRRL) team at the Oakland University Eye Research Institute has been awarded two significant grant funding awards: a National Institutes of Health Small Business Technology Transfer grant and a grant from the Michigan Emerging Technologies Fund. The PRRL team will use the grant funding for one year to investigate the ocular therapeutic drug Noregen™, which may promote regeneration and repair of damaged retinal blood vessels in the human eye. For an update on how the funds raised last year further impacted the research initiative, see “On the Research Front” below.

It was banner year from the PRRF community perspective as well:

- Our community numbers ~1500 strong and growing.
- Ongoing content development continues to add value to the PRRF website. We are particularly proud of the educational advocacy initiative on the site – a top priority project undertaken in response to the feedback we’ve received from the PRRF community. This issues installment in the “Focus on the Family” section by Board member Chuck Walls and his wife Donna speaks poignantly to the impact raising a low vision child has on the rest of the family.
- We expanded our social media initiative under the leadership of Courtney Birchmeier. Courtney created a new Facebook page to post events and other information (https://www.facebook.com/pediatricrrf/) in the interest of facilitating communication with and within the PRRF community.

We’ve gotten a lot done!

As this very successful year comes to a close, we undertake our 2019 year-end initiative to purchase a hand-held pediatric Optical Coherence Tomography, (OCT) device for use in the operating suites. This device allows for noninvasive evaluation of the anatomy of the eye over time, facilitating informed decisions regarding the need for surgery. We are also working on an informational conference with both a science component and a community-based component for the late spring of 2020. Your support is invaluable in reaching these goals.

To close, a very special “thank you” to the leadership of the PRRF Board, the enthusiastic dedication of our PRRF volunteer community, and to our many generous contributors.
From all of us on the Board of the PRRF, best wishes to you and yours for happy holidays and a peaceful and prosperous New Year.

Sincerely,

[Signature]

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

Hope For Vision Walk Fundraiser Update

Total funds raised for this event were $21,000
This fall the Pediatric Retinal Research Foundation (PRRF) held its inaugural Hope for Vision Walk which took place on September 28th in Bloomfield Hills, MI. The PRRF’s mission is to support the community of families impacted by blinding pediatric retinal diseases and the quest for a cure. The walk included music, refreshments, raffles, a Zumba warm-up, and activities for kids to enjoy.

In total 12 teams (95 people) joined together and walked the neighborhood of Bloomfield Hills for 1.5 miles. Teams included children and adults and their families affected by pediatric retinal diseases, local foundations, local practices, and numerous groups showing support for this cause. The walk was promoted on social media, in local businesses, and by word of mouth for weeks receiving a significant amount of donations for the PRRF’s critical work. Total funds raised for this event were $21,000.

The ARC team included 31 ARC staff and their friends/family members. The ARC team accounted for $5,800 of the total amount raised which was more than 5 times the initial goal set for the team! Team members promoted the walk on their social media nearly every day for many weeks and many were able to raise at least $100, which was a personal goal for everyone on the team. Additionally, many ARC staff members volunteered for this event, which was a huge help. It is truly amazing to see the dedication ARC staff members have to the ongoing research and support the Pediatric Retinal Research Foundation provides families. Everyone is so excited for next year’s walk!

~ Written by Tracy Martin and Photography by Tom Treuter
**On the Research Front**

**DNA Sequencing Process Developed in the PRRL at Oakland University Successfully Activated this Summer.**

A special thank you to all of our donors who helped to buy the new sequencer and allow for the continued success of our Biobank! We thought an update on the genetics might interest anyone who has ever wondered why genetics are so important in advancing new therapies.

Everyone’s DNA sequence contains thousands of variations. Most of these do not affect the proteins they encode or produce; however, some may cause (or exacerbate) disease. The Pediatric Retinal Research Laboratory analyzes the DNA sequences of patients and their relatives to help decipher which variations are responsible for Wnt-associated vitreoretinopathies (FEVR, Norrie Disease, Osteoporosis Pseudoglioma Syndrome) and Congenital X-linked Retinoschisis. Thanks to our supporters, this work has been accelerated by the recent acquisition of a Next Generation Sequencer called the I-Seq. The I-Seq allows us to sequence all known candidate genes (8 genes) in up to 40 patients during one run! Up until this time, using conventional sequencers, only a small portion of individual genes could be analyzed per run. What took years with the older instrument, can be run in a day on the I-Seq! The benefit of this new technology includes speed and a dramatic reduction in cost from several thousands of dollars for 2-3 genes to less than $250 for 8 genes.

We have already run the DNA sequencing of 32 subjects using the I-Seq and are in the process of analyzing the data. One way we can do this is to compare the sequences of parents and a child with a specific disease, such as FEVR. In this method, we look for variations that an affected parent and the child with FEVR have in common but that the unaffected parent does not share. Another method is to group patients with a given variation and assess if there is a correlation with disease severity. We expect that this data will help to guide development of new treatments. Furthermore, the sequencing project methods can be shared to help other groups who wish to design custom sequencing panels for other kinds of rare genetic conditions.

Most exciting is that the results will have immediate implications on disease management. Physicians will be able to use this information to more closely monitor patients with "severe" variations and getting ahead of flare-ups. In short, the I-Seq has brought us much closer to our goal of having a treatment for our patients with FEVR and similar vitreoretinopathies.

We again would like to thank our supporters and the team for all of their dedication, efforts, and hard work.

**The Team:**
Wendy Dailey, BS  
Ken Mitton, PhD  
Kim Drenser, MD, PhD  
Pediatric Retinal Research Lab  
Eye Research Institute  
Oakland University  
[http://www.oakland.edu/eri](http://www.oakland.edu/eri)

~ Written by Kim Drenser, MD, PhD

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**On a Personal Note**

A Young Philanthropist
Rebecca Shamah turned 12 this past June and began to prepare for her Bat Mitzvah. In keeping with her family’s philanthropic values, she decided to use the Bat Mitzvah as a platform to raise awareness as well as money for a cause dear to her family - research to help children who have been born without sight.

Rebecca’s cousin, Isaac, was born blind just over two years ago. His vision impairment is a result of a form of FEVR, and he has been treated by Dr Capone since he was a tiny baby. Rebecca is very close to her cousin and began to think of how she could help others see Isaac’s journey and become more understanding of the challenges that anyone with vision impairment faces.

In preparation for the Bat Mitzvah, Rebecca and her family set up a Go Fund Me page for VRRF, which raised almost $10,000. At the actual Bat Mitzvah service, Rebecca wanted the 200 attendees to also physically understand how it is to be blind. During Rebecca’s speech, the guests all put on blindfolds and listened while Rebecca talked about her special bond with Isaac as well as the challenges that he and other children who cannot see face. She spoke of the hope that the research being done now holds for preventing or improving children’s vision problems in the future.

At the party which followed, the entire hall was set up with black lights which showed what it is like to move in the world of darkness. Rebecca designed a logo of an eye to focus all on vision and how important it is. Everyone had fun but many were deeply touched by the new personal knowledge of how the world without sight is challenging.

Rebecca is a wonderful example of how a caring group can make an impact. The financial donations and the awareness which were raised are very meaningful contributions to the work of the Pediatric Retinal Foundation.

Thank you, Rebecca, and many thanks to her family and friends.

~ Written by Kay White

~ Photograph courtesy of Rebecca Shamah

Focus on the Family

Family Dynamics with Low Vision Special Needs
They call it the “after the diagnosis” phase. Still to this day, I remember the sinking feeling I had in my stomach when we were first told about our son’s diagnosis and the explanation that followed. That was twelve years ago. I vividly remember it all: the confusion, the frustration, and the feeling of helplessness. I remember panicking about what to do. We felt alone.

I wanted to write this article to share the many sources of support and services that are available for your children and families of children who are visually impaired. You are NOT alone! There are other parents and families out there who want to connect and partner with you as you start this new journey with your child and with your family.

The realization of visual impairment for your child is a shocking event for you and your family no matter what age of diagnosis. It affects all aspects of your entire family on personal and financial levels. Marriages fall apart because one or both of the parents become so hyper-focused on treatments, hopes for a cure, or even the daily tasks of meeting their child's needs that they forget they are a married couple. We went years without doing anything special except focusing on our son’s disability. Find time for each other!

This also applies to the other family members, specifically the siblings. The amount of time and energy that parents spend with their visually impaired child is huge and siblings can easily feel forgotten. This can foster unintended bitterness or jealousy and siblings may turn to other ways of getting attention. The spectrum is wide – from “acting out” to grab additional attention to reclusion and depressed feelings and require significant parenting on top of the worries and concerns for the special-needs child. Look out for these changes and face them head on.

Lastly, financial impacts within a special needs family may exacerbate the family dynamics. Whether it is a financial “tightening of the belt” across the board or specific focus on necessary costs for the special need, the additional stress can be huge. Teamwork throughout all, including from the impaired child, is necessary and remember to take the time to thank each other for even the little things.

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