IMPORTANT NOTICE

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Letter from the President

Within the universe of conditions, we tend to as pediatric retina specialists, Norrie Disease (ND) is among the rarest. Most retina specialists will see 2-4 such children over a 30+ year career. If one has a child with ND, it's near certain that there is no-one else in his/her hometown with a family member with this condition unless he/she lives in a large metropolitan area. The NDA has taken shape to address the need for community – a "place" for parents and others impacted by this condition share information and resources. In this, the NDA and our own PRRF are highly aligned.

Over the years I've held that I often learn more than I teach when attending a conference as an invited speaker – as was the case again at the recent 4th annual Norrie Disease Association (NDA) meeting in Boston on August 9-11. This year’s meeting was attended by several members of our own community (see the image below). At this meeting I learned of two high priority issues for the ND community.

Most males with ND experience hearing impairment as well as vision loss, and half have developmental/cognitive issues as well. Vision limitations declare themselves at birth or shortly thereafter, and the cognitive issues are what they are. But the parents watch their sons lose their hearing gradually, and concomitant further sensory isolation, typically starting in the second decade of life. Though I've been tending to boys and men with Norrie disease for more than 20 years and am quite aware that hearing loss occurs, I found the importance of this issue to the ND community striking.

The other high value issue for the ND community is collaboration between investigators. Predictably, there is a high level of interest in scientific advances relating to Norrie disease and its manifestations. In particular, members of the ND community want from clinicians and scientists that we share ideas and information, recognizing correctly that advances proceed more quickly as a consequence of collaboration. Notwithstanding the challenges to an open and collaborative scientific world – publication pressures, competition for funding, and more – it is reasonable to expect that we clinicians and scientists rise to that...
expectation. In our own pediatric retina practice we have had an open-door policy for decades for that very reason. At any time, there are 3 visiting scholars interested in pediatric retina drawn from all over the world, and we are booked well over a year in advance. Our philosophy is that what we have learned of the pediatric retinal conditions we tend to doesn’t belong to us – it belongs to the children. Ours is but to share it.

PRRF exists to research pediatric retinal diseases and support the community of families impacted by these conditions - your help enables us to continue to offer opportunities such as the ones to investigate treatments for ND and similar retinal conditions.

Antonio Capone, Jr., MD, President

Team Photo from the recent NDA meeting. From L to R: Alison and Ted with their son Nate, Stephanie, Dr. C, Carmary and Robert with their son Robert, Heather and Matt with their sons Mackenzie and Kaelen, and Chris and Kiah.

On the Research Front
A primary research focus in the Pediatric Retinal Research Laboratory (PRRL) is to understand how the eye develops and to use this knowledge to create new therapies for blinding diseases. In the retina, many eye disorders result in abnormal blood vessels. The blood vessels within the retina have a very specific structure to allow them to transport blood and nutrients to tissues without “leaking” along the pathway. These are known as "non-fenestrated" blood vessels. Non-fenestrated blood vessels limit vessel wall leakiness by having tight junctions between the cells that form the vessel walls. We have been working on a therapy that strengthens tight junctions by turning on the production of the proteins responsible for this barrier. Vascular endothelial growth factor (VEGF) is a protein that, when overexpressed, results in the breakdown of tight junctions, resulting in unwanted leakage of serum, lipids, and cytokines into the retina. This leakage results in swelling which in turn decreases vision. This occurs in many retinal diseases, such as Familial Exudative Vitreoretinopathy (FEVR) and Retinopathy of Prematurity (ROP). This figure demonstrates the ability to reverse this pathology by turning up the production of claudin, one of the primary proteins which reinforce tight junctions.
On A Personal Note

After having a healthy pregnancy and delivery, our world was turned upside down when our daughter, Presley, was diagnosed with Stage 5B FEVR (bilateral retinal detachment) a few days after she was born and was completely blind.

During her first week of life, we saw multiple doctors in Texas who either did not feel comfortable operating on her given her age or did not feel surgery was even an option due to the severity of the presentation in her eyes. We were told her eyes would not grow so they would always be small and because she was already the most severe stage, nothing in her eyes would change. Heartbroken and searching for any sliver of hope, it became clear to us that we needed to travel to Detroit to see one of the “FEVR experts,” Dr. Trese or Dr. Capone.

After our initial visit with Dr. Capone a few weeks later, we knew we were right where we needed to be. He has been a godsend to us both medically and emotionally and was the first person who gave us hope. He has taken the time to thoroughly educate us on this rare disease and allow us to ask as many questions as we need.

We made three trips to Detroit before Presley turned one for three rounds of surgeries on each eye. While we initially were told her eyes would not grow, when we went to see Dr. Capone, her eyes had gone from being small for her age to being huge due to fluid buildup in her eyes. He saved her from getting Glaucoma by relieving pressure and protecting her optic nerves. Dr. Capone had to remove both lenses and more and more of her iris with each surgery. When he went in for the second surgery on the first eye, he thought we had won the battle but lost the war. Her retina was sinking back slowly like we had hoped but there was no blood flow to the retina, meaning the retina was underdeveloped. Instead of stopping there, Dr. Capone kept going. After removing more and more scar tissue and draining dried blood, blood vessels finally appeared. Dr. Capone does not give up until it is not safe to continue and always has the child’s best interest in mind. His commitment and passion to help these kids, who often times do not have a positive prognosis, is something that truly leaves me speechless.

Managing your expectations makes the smallest things feel like the biggest victories. Finding a doctor like Dr. Capone was a victory. Draining the fluid before she got Glaucoma and her optic nerve was damaged was another victory. Regardless of any light or vision we may or may not get, we are at peace with that.

Now two years old, Presley is a sweet, happy, and very chatty toddler. Her auditory processing and auditory memory is incredible. She has been learning how to walk with a cane and will soon begin learning Braille. She amazes us every day with how easily she picks things up and adapts. While we would give anything for Presley to be able to see, she has given us a new perspective on life and changed our world for the better. We are so thankful for Dr. Capone and the work that PRRF is doing to help families like us.

Caroline and Michael Halbert
Venkat N. Reddy, Ph.D., a world-renowned vision researcher and recognized expert on the metabolism of the lens and the formation of cataract, died peacefully on June 30, 2018 in Rochester Hills, Michigan at the age of 95. Dr. Reddy was an original member of the Board of Directors of the ROPARD (Retinopathy of Prematurity and Related Diseases) Foundation, which was established in 1990 and preceded the Pediatric Retinal Research Foundation.

Dr. Reddy grew up in the small village of Chintakunta in India, which is now part of the city of Hyderabad, as the youngest of four sons. In 1945, he received the B.Sc. degree in Chemistry from the University of Madras, and encouraged and supported by his brother, Dr. Narisimha Reddy, he emigrated to the U.S. to obtain the Masters and PhD degrees in Biochemistry from Fordham University. This was followed by a four-year post-doctoral fellowship at Columbia University and a brief stay as a research fellow at the Banting and Best Institute in Toronto, Canada. In 1956, he joined the Kresge Eye Institute at Wayne State University in Detroit, Michigan as a faculty member, teaming with Dr. V. Everett Kinsey for what would be a marvelous 20-year relationship. In 1968, the two investigators left the Kresge Eye Institute to establish the Eye Research Institute (ERI) at Oakland University in Rochester, Michigan. Dr. Reddy served as ERI Director from 1975 to 1997, and under his leadership, the Institute gained national prominence as a leading ophthalmic research center. Retiring from Oakland University in 1998, Dr. Reddy joined the faculty of the University of Michigan Department of Ophthalmology and Visual Sciences as a Senior Research Professor. He ended his remarkable 50-year vision research career in 2006, returning to Oakland University as a Distinguished Professor of Biomedical Sciences, Emeritus to interact almost daily with ERI faculty, staff and student researchers for another decade.

Early in his career, Dr. Reddy collaborated closely with Dr. Kinsey to conduct pioneering studies on the chemistry of aqueous humor. Many of the transport mechanisms that contribute to aqueous humor formation were elucidated by this work. A major part of Dr. Reddy’s research effort was the careful, detailed analysis of steady state levels of free amino acids, electrolytes and related compounds in the aqueous humor, coupled with the determination of the mechanisms of transport of these molecules into the various compartments of the eye. This work is considered a major contribution to our understanding of lens physiology and resulted in Dr. Reddy receiving the prestigious Friedenwald Award in Ophthalmology in 1979. In addition to his investigation of aqueous humor dynamics, Dr. Reddy is also well known for his extensive studies on the degradation and synthesis of reduced glutathione, the major antioxidant compound present in the lens. His numerous research accomplishments spanned both the lens and glaucoma fields.

Dr. Reddy’s research was continuously supported by the National Eye Institute (NEI), NIH for more than four decades, and included a coveted ten-year MERIT Award received in 1989. He published over 180 peer-reviewed articles during his career and received many research accolades including the Alcon Research Recognition Award (1984), Michigan Scientist of the Year Award (1991), and Oakland University Research Excellence Award (1993). On the occasion of his 90th birthday, the LV Prasad Eye Institute in Hyderabad sent him a beautiful glass sculpture created by the Indian artist Sisir Sahana for “his outstanding efforts in promoting vision research in India”. Dr. Reddy had close connections with leading vision researchers from all over the world. Each year for 25 years, he invited a few dozen of them to attend a “Biochemistry of the Eye Conference” held in Meadowbrook Hall (now a National Historic Landmark) at Oakland University to present results of their most recent studies involving all tissues of the eye. Dr. Reddy and his wife Alvira had a special fondness for Japan. In 1983, he was invited by the Japanese Ophthalmological Society to present its Guest of Honor Lecture, an honor not usually accorded to basic scientists. Beginning in 1980, Dr. Reddy invited over 20 visiting scientists from Japan to spend two years each working with him in the ERI. One result of these highly productive collaborations was the creation of an immortalized human lens epithelial cell line (SRA 01/04) that is currently being used by researchers all over the world to investigate lens epithelial cell metabolism.

Dr. Reddy was also a well-recognized leader and advocate for vision research outside of the laboratory. He served on the editorial boards of Investigative Ophthalmology and Visual Science and Experimental Eye Research for many years and was a frequent member of NIH Study Sections. In the 1970’s and 80’s, he served on the NEI Board of Scientific Counselors (1977-81) and the National Advisory Eye Council (1982-87). In 1986, he was elected as President of the Association for Research in Vision and Ophthalmology (ARVO) and served as President of the National Foundation of Eye Research from 1984 until his passing.

In 1998, over 50 of Dr. Reddy’s colleagues attended the Venkat Reddy International Symposium at the Airlie Conference Center in Virginia to thank him for his many years of leadership and dedication to ophthalmic research, and to honor him for his life-time achievements (see Reddy VN, A forty-two-year voyage through vision research, J. Ocular Pharmacol., 2000). He will be remembered as an excellent scientist and academic leader, a true gentleman scholar and a key figure in the founding of ophthalmic research as a respected discipline. He was preceded in death by his wife of 57 years, Alvira. He is survived by his loving son Vinay (Laura Smidchens), daughter Marlita Reddy-Hjelmfelt (Eric Hjelmfelt), and 17 nieces and nephews.

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