

Happy Anniversary



THE VISION of CHILDREN



This year we celebrate two decades of milestones and successes in our tireless effort to complete our Mission, "To cure hereditary childhood blindness and vision disorders, and to improve the quality of life of visually impaired individuals and their families." This Mission has guided our research, funding, scholarships, and computer monitor programs for the visually impaired. Twenty years ago, genetic research into ocular albinism(OA) didn't even exist. Now, the VOC supports 23 researchers at 12 institutions worldwide, and helps fund studies for over 25 associated genetic eye disorders. That's progress, and that's success!

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Happy 20th Anniversary VOC!

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THE FOREFRONT OF RESEARCH

Over the years we've channeled the majority of our resources into genetic research, pushing the known science into new frontiers. For example, in 2001 the VOC reported that, thanks in part to its funding,



OA gene testing for humans became available. Offered at the Baylor College of Medicine in the US, and Laboratorio Biologia Molecolare in Europe, the testing allowed mothers to learn if there was a risk of passing the OA condition to their children, and helped determine if a patient with inconclusive physical symptoms had OA. In 2006, the VOC funded testing for the first

electrophysiological study of individuals with OAI. This non-invasive work explored the misrouting from optic nerves to the brain. And in 2008, Professor Robin Ali's team at the University of London's Moorfields Eye Hospital conducted pioneering research resulting in the world's first gene therapy trial for a vision disorder. Additional trials took place at the Children's Hospital of Philadelphia and the University of Florida in Gainesville. Most patients reported significant improvement in their vision, and researchers found that treated eyes became approximately three times more sensitive to light. This was a major milestone because gene therapy has applications for all genetically caused vision disorders. This important gene therapy trial proved the efficacy of the pathway to success that VOC embarked upon two decades ago.

We are proud to have played a significant role in these important advances. The VOC has positioned itself at the forefront of genetic eye disorder research and has helped create the conditions that have pushed the scientific field forward.

WORLD SYMPOSIUMS

Beginning in 2000, the VOC has facilitated research collaboration and sharing among scientists from around the globe by hosting the World Symposium on Ocular Albinism. The 7th, and most recent, was held in 2009. Each conference is an open forum for invited scientists, at some of the world's finest research institutions to learn firsthand about each others' studies through presentations and Q&A. The



Symposium encourages new directions in research, the creation of new research partnerships, and raises the all-important awareness that generates funding for new studies.

STELLAR PROGRAMS

Over the years, we've not only facilitated significant advances in science that are moving us closer to a cure, but have initiated programs that are touching the lives of people with vision disorders every day. Our computer monitor system (CMS), started in 2003, delivered a profound impact to children with OA from a simple idea. Our founding Chairman noticed that his son, who has OA, was having great difficulty in learning how to properly use the computer and enter information by typing. He had to lean over the keyboard to see the monitor closely, which impeded his ability to type and learn. By creating a system to move the monitor to the student, both problems were solved and thousands of visually impaired children have benefitted dramatically from this invention. The VOC has given these systems to schools, libraries and needy children throughout the nation. Students using the CMS reported improvement in their typing skills by 50-85 percent! This year, we are looking to new technologies (see: Pocket Viewer Article), and we're very excited! We're still offering the book, "All Children Have Different Eyes," expanding our Family Network, which enables parents to contact families facing similar challenges in their geographic region, and much more! Please consider joining our network or helping us to help others in need by registering online at www.visionofchildren.org.



THE HEART OF THE MATTER

These stories are reflective of the amazing successes and accomplishments of our talented team, but they are only half the story; there's another way to describe what the VOC does, what it has accomplished, and why it's all so important.

It's people. It's families, children, and parents. It's lives that we've touched. At the end of the day, when it's all said and done, helping people is what the VOC is about. When Sam and Vivian Hardage's infant son was diagnosed with OA nearly two decades ago, Sam and Vivian founded the Vision of Children-above all else-as concerned parents. Since then, as our Family Network has grown, we've been able to give answers and hope to thousands of other parents and individuals. Back in 2001, we wrote about Judith and her three week old son Joe. She and her husband first noticed a continuous back and forth movement in Joe's eyes when he was three weeks old. Their search for answers was hampered by a lack of information and insufficient diagnoses. She described a trip to the pediatric ophthalmologist as the absolute darkest day of their lives; a cursory examination was followed by a pronouncement of oculocutaneous albinism, a prognosis that Joe's eyes could probably not be corrected better than 20/200 - 20/400, and that nothing could be done to help him. All Judith and her husband could see was a life of limitations for their child.



Tommy Saker (from left), Christine Saker, Sherri Saker, Joe Saker, Richard Saker, Laura Saker

But their search for answers led them to the Vision of Children. VOC put them in contact with Dr. Richard Lewis, a foremost expert in OA (and a member of the VOC's Scientific Board of Advisors). He answered their questions, dispelled misconceptions, and made recommendations. After it was all said and done, Judith was able to say with the happiness and relief of a loving parent, that their pediatrician was able to give them wonderful news; if Joe, 8 months old, wasn't wearing glasses, they wouldn't know he had any vision problems!

This story has repeated itself over the years: a mother of two children with OA who found support in sharing her experiences with other parents over our Family Network, a 16 year old son with a retinal degenerative disorder who was given low-vision monitor equipment from the VOC. The details vary, as do the endings, but the common thread is hope. To love someone and not know how to help them is a terrible feeling. For twenty years, the VOC has worked to provide hope to people with vision disorders through answers, resources, support, and ultimately, a cure. To everyone we've helped, and everyone we've yet to give our support, we pledge to persevere until all genetic vision disorders are cured!

The VOC would like to extend a special thank you to all our supporters over the last 20 years. From the one time donor, to fundraisers of all ages, to our major sponsors, to all our supporters, to corporate givers, and to the countless volunteers who have donated their time and resources- we are profoundly grateful! 🙏



THE PATHOGENESIS OF OCULAR ALBINISM TYPE 1

Biomedical research is referred as “basic” when it is mainly aimed at enhancing our knowledge of biological systems in health and disease, without directly concerning the development of diagnostic or therapeutic protocols for patients. Understanding basic mechanisms responsible for human diseases and unraveling why and how a disease develops, is an essential and obligatory step required for any subsequent attempt to create diagnostic procedures, as well as identifying and evaluating the efficacy of possible therapies.

In genetic disorders such as albinism, the process leading to an affected individual is primarily caused by a mutation in the genomic DNA, which in turn results in a series of successive alterations at different levels in the organism. This is collectively defined as the disease pathogenesis. Thus, genetic mutations in the genes for albinism generate defects in the pigmented cells of the skin and eyes, leading to an abnormal development of the retina and optic nerves, which results in visual impairment affecting persons with albinism. As underlined in the title of the Seventh World Symposium on Ocular Albinism held in San Diego last Spring, “Understanding the Pathway – Discovering a Cure”, it is very important to study in detail all these intermediate steps leading to a disease, because each of them could provide chances for diagnosis, prevention, or therapy.

As most genes implicated in albinism, the gene primarily responsible for ocular albinism type 1 (OA1), is already known and its mutations have been characterized. Therefore, the target of our research laboratory in Milan, Italy, is to understand the pathogenesis of ocular albinism at the next step in the pathway, which is at the cellular level. We want to unravel why loss of OA1 function leads to abnormal development of the melanin-containing organelles, named melanosomes, within the pigment cells of the skin and eyes. In fact, melanosomes are bigger and less numerous in persons with ocular albinism, and these intracellular defects may determine the retinal abnormalities responsible for poor vision. To this aim we have developed melanocyte cell lines from an OA1 mouse model and found that, in addition to regulating the formation and the size of melanosomes, OA1 also regulates their transport and motility within the cells. Thus, our results suggest that ocular albinism might indeed result from a different pathogenetic mechanism than previously thought.

We are now trying to define the molecular mechanisms by which OA1 accomplishes its function, including its activation mechanism and downstream effectors in the signaling cascade. Our goal is to be able to dissect its intracellular network of interactions in the near future. ☺

EVIDENCE-BASED MEDICINE FOR THE TREATMENT OF INFANTILE NYSTAGMUS

In patients with infantile nystagmus syndrome (INS), the continuous movement (‘jittering’) of the eyes causes the image of the outside world to be blurred, which typically reduces visual acuity. INS may occur by itself or in combination with other conditions that stand in the way of the normal development of vision, such as albinism. If nystagmus is present during the first years of life, this ‘deprivation’ may also lead to long-lasting poor vision.

A relatively new surgical approach to improve the eye movements has shown beneficial results in adults and older children. And even though the improvements in visual acuity were sometimes very modest, it is promising for young children and infants because earlier surgery may have the additional benefit of reversing or preventing

long-lasting vision loss.

With a new technique for analyzing children’s eye movements, developed by The Retina Foundation of the Southwest with grant support from The Vision of Children, we are now underway to evaluate the effects of nystagmus surgery in children in the age range of 3 to 10 years. By monitoring eye movements as well as visual acuity over time, we hope to gain valuable knowledge about vision loss in children with INS and whether vision loss can be reversed. We currently have enrolled 12 children in whom INS appears to have occurred by itself, as well as 10 patients in whom INS is associated with albinism, 4 patients with bilateral optic nerve hypoplasia, and 6 patients with a variety of these conditions. All of these children either have undergone treatment or treatment is planned. We look forward to communicating the first results by the end of this year. ☺



Joost Felijs, PhD
Retina Foundation
of the Southwest

TAKE ACTION

DOCTOR MEMBERSHIP

The relationship between a patient and doctor is really quite simple; one person seeks the help and support that the other has dedicated their life to providing. To that end, we would like to highlight our new Doctor Membership Program – a great way for doctors to connect their patients with VOC’s vision resources and support, while at the same time, growing their practice.

The Vision of Children Foundation has 20 years of experience in areas pertaining to genetic eye disorders: pioneering research, international networking, physical and software solutions, and social support. When a doctor becomes a “Vision, Passion, Hope, or Cure” level member, they are effectively expanding their own ability to reach those patients in need of help. Their ability to make-and opportunities to receive-recommendations increases, as do immediately available resources, such as brochures, bookmarks, newsletters, and the book, “All Children Have Different Eyes.” The more families and individuals learn about us, the more frequently they’ll approach their doctors with questions and referrals, and that means more people with genetic eye disorders will get the treatment and support that they need. Our Family



Network (FN) connects families and individuals around the globe, and we want to be able to give current and future FN members more opportunities to receive the best care available. Linking doctors to the VOC through our Doctor Membership Program is an important step in giving people access to VOC’s resources and support.

At a minimum, member doctors receive the informational materials for their waiting room, use of the VOC logo on their personal website, recognition of membership on the VOC website, and more. Together, with Member Doctors, we can provide help and support to people with genetic eye disorders everywhere.

To learn more about joining our Dr. Membership Program, please visit our website (www.visionofchildren.org) or call our office at (858)-799-0745. ☺

KID OF THE MONTH



Once again, it’s time for Kids of the Month. This section began as Kid of the Month, singular, but there are just so many great young people doing great things that we had to expand! We’d like to introduce Sophia McClellan,

her sisters Kate and Molly, and their cousins Madison and Megan. They are lemonade stand fundraisers extraordinaire!

Sophia is almost 9 years old, but she was just 6 when her mother brought home a Vision of Children flyer from a ShopRite grocery store. Sophia had been asking about helping out a charity (Sophie has amblyopia, and she and her younger sister Kate both wear glasses), and together they decided that the VOC would be perfect! So, that summer, Sophia and Kate set up a lemonade stand at the “Dog Day Road Race,” a 5-mile foot race with lots of thirsty runners. At

the end of the day, they had raised \$15 for the VOC! But that’s not the end of the story. Over the winter, the two put together handmade barrettes with buttons and other decorative items. Then, the next summer, they combined the lemonade stand with their barrettes in a tour de force of junior fundraising! This time they were joined by their youngest sister Molly, and cousins Madison and Megan. They raised \$85! Very impressive, and very much appreciated!



We were told that they hope to make this an annual event, and we hope so too. The VOC gives a big “Thank You” to warm-hearted Sophia, Kate, Molly, Madison, and Megan. Maybe Forbes magazine will want to interview these kind hearted young entrepreneurs! ☺

FAMILY NETWORK

POCKET VIEWER

Those of you who have been following the VOC are probably familiar with our Computer Monitor System(CMS) program. Since 2003, we've provided specialized viewing systems for maximizing educational opportunities for low vision students with great success. This year, the VOC is rolling out the red carpet for a different kind of technology; handheld video magnifiers, or, pocket viewers. We will be using GW Micro's "Portable SenseView Duo."

Unlike computer monitor systems, the SenseView is an aid that students and other users can carry with them anywhere. It's about as long as a dollar bill, and fits easily in a backpack or small bag. The SenseView's purpose, via a screen and camera, is to magnify and/or digitally enhance images of text, handwriting, signs, presentations, and whatever else its low vision user is interested in examining. The device represents a huge benefit to everyone from students who have difficulty following their lesson on the whiteboard, to individuals

trying to make out the address on a building across the street.

Part of our mission has always been to "improve the quality of life of visually impaired individuals..." and we believe this handheld device will help us accomplish that goal. It will help free low vision students from the burden of listening and memorizing when unable to see clearly, and give them the opportunity to learn by seeing more clearly.

If you are interested in learning more about this device, please go our website, www.visionofchildren.org or call our office at (858) 799-0810. ☺



SPECIAL THANKS

The Vision of Children Foundation would like to recognize the outstanding support of Jordan and Mack Portnoy, who picked the VOC as the recipient of their school fundraiser, and helped raise awareness and funding for our cause. Congratulations to you both on your recent Bar/ Bat Mitzvah, and thank you again for your philanthropic efforts on behalf of the Vision of Children Foundation. We would also like to congratulate Kevin Graff on his Bar Mitzvah, and specially thank him for selecting the Vision of Children Foundation as the beneficiary for his Bar Mitzvah. We would not have had the pleasure of knowing the Graffs if it

were not for George Triebenbacher, so thank you, George, for introducing us to these wonderful people, and for your continued longtime support of the VOC. A final thanks goes out to Marissa Makram for her amazing work in organizing "A Night For A Cure." This event was a huge success and generated significant funding and awareness for our cause. Thank you to everyone for your dedication and support of the Vision of Children Foundation; because of people like you, we are steps closer to reaching our mission and finding a cure for childhood blindness and vision disorders! Thank you! ☺

PAST EVENTS



*Jordan and Mack Portnoy
(from left)*



Kevin Graff



Joseph Makram