



In the SPOTLIGHT Focusing on Researchers and Kids Alike



Moving forward we will regularly turn the spotlight on both our researchers and our Family Network Members. For each spotlight we hope to provide a short video, which we will post on our Web page and YouTube channel. However, it can also be written words with a photo, which we will post on our Web site or in our blog (or the newsletter!). The researchers will share how they became involved in vision research and what they are currently working on, as Vittoria does in this issue. Our members will share their journey with low vision and what Vision of Children's work means to them, as Riley did this May. If you or your child would like to be featured, please let us know! It will be especially fun to highlight those who were previously written about who are now young adults. The videos do not have to be professionally made and can be as short as 2-minutes or as long as 6-minutes.

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Meet Vittoria Schiaffino Steadfast VOC Researcher

I am from Padua, a medium-sized town close to Venice in the north-east part of Italy. I did all of my undergraduate school and attended university there. Although I studied medicine, my original idea was always to become a researcher because I didn't like the hospital environment, but was rather fascinated by labs and experiments, and by DNA in particular. In Padua, I also had the chance to cultivate my other great passion, mountain sports: ski, alpinism, climbing in the Dolomites and Alps nearby (pictured, above). After obtaining my MD degree I moved to flat (!) Houston, TX, to Andrea Ballabio's lab to work on human genetics. At the time I didn't know much about albinism, so that, considering my passion for the mountains, Andrea always tells a funny story about the first time we met: he says that when he asked me if I wanted to work on albinism I replied: What?? Ahh, alpinism! Oh, YES YES!!!

Of course it is a joke, although in reality I began working on albinism more or less by chance, since at the time Andrea's lab was searching for disease genes mapping along the X chromosome and one of the projects regarded the search for the gene responsible for Ocular Albinism (OA). Since only one person was dedicated to that project and Andrea wanted to have a stronger team, when I landed in Houston he placed me on OA. From then on (it was 1993) I continued to work mainly on ocular albinism, first of all by identifying the responsible gene, OA1, together with Andrea and my colleague MT Bassi.

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8th World Symposium on Vision Disorders to be Held in San Diego

Once again, we will gather vision researchers from around the world to discuss their research and share all of the exciting advancements that are being made. Family Network Members will also be welcome for a special session dedicated to you and answering any questions you might have.

The exact location has not yet been determined, but it will be held in San Diego November 6 - 8, 2013, so mark your calendars!



National Grocers Association Recognizes ShopRite's Support of VOC

We shared in the fall newsletter that the six year total of the Saker ShopRite contributions to VOC has reached nearly \$1 million. We felt it news worthy, so we sent out a press release—and the National Grocers Association (NGA) agreed! NGA included the announcement and associated press release in their March 13th "Express Lane" newsletter that is sent to all of their members. It's our hope to garner support from more NGA members in our continuing fundraising efforts.

San Diego Area Grocery Stores Support us Too!

We now have a couple of grocery stores in San Diego County (and more potentially) raising money at their registers for Vision of Children. Wally's Market in Imperial Beach has VOC donation boxes on the counter at each register where their customers are showing their support by dropping in donations. Stump's Market has two locations: the Rancho Santa Fe Village Market supported us during the month of April and will again in October and the Point Loma Marketplace has dedicated April and May to VOC this year. Please be sure to visit these markets when you are in these areas and thank them for supporting us! Pictured below, Vanda from Point Loma Stumps (left photo) and from Wally's, May (on left) and Sam.



Team VOC - Racing for Research Running Strong

It is remarkable how much support each of our runners has received from their friends and family. Family Network Member, Cyndie Schade (pictured right, top), ran the San Diego half marathon this March and had this to say after raising close to \$500, "I have awesome friends and family. It's very humbling to say the least. So happy I'm doing this!" Ultimately, Cyndie raised \$3,140— 5% over her goal—and she did it by sending e-mail messages to family and friends and posting about it regularly on her Facebook page. People clearly want to help and are even more motivated when they can help an organization that is trying to support a friend or family member.

Team VOC was created to give our supporters a way to fund-raise for VOC while doing something they might already be doing (or might be inspired to do) and the results have been astonishing! A wonderful by-product is that it is giving us exposure and extending our reach to new families with children with visual impairments, while informing more people about the important work we are doing.

A very strong spring: We had seven people who work for the Montage Hotel, Beverly Hills (pictured below), run the LA Marathon in VOC's honor and they raised more than \$2,200! Family Network Member, Ashly Stohl (pictured right in the VOC tank - before & after), ran the "Run or Dye" 5K in LA with two friends recently and raised more than \$2,500 in less than ONE week. Amazing! Speaking of amazing, we briefly mentioned George Triebenbacher's Team VOC participation in the fall newsletter. The NYC marathon was cancelled due to Superstorm Sandy, so he did not run, but raised more than \$18K—he has indicated he'll run the NYC Marathon twice this year to make up for his missed miles! :-)



TEAM VOC EVENTS COMING UP

TWIN CITIES: We are a "Participating Charity" in the Twin Cities Marathon, happening October 6th, 2013. We need runners! We also need volunteers, so if you or anyone you know lives within driving distance of the Twin Cities, please let us know and we will let you know how to get involved.

LONG BEACH: There are a few people who are running the Long Beach marathon or half marathon and will be raising money for VOC in the process. Would you like to join in the fun on Oct. 13?

Check out our CrowdRise page for all of these race fundraising pages: <http://www.crowdrise.com/visionofchildren>

RESEARCH UPDATES

Oculocutaneous Albinism (OCA) – Human Clinical Trial Underway Vision Response to Dopamine Replacement Trial

The Vision of Children has been funding research by Michael Struck (pictured right), MD, a researcher based at the University of Wisconsin, Madison, since the fourth quarter of 2012. He recently started recruiting patients for a human clinical trial to determine if there is an improvement in vision in response to dopamine in individuals with oculocutaneous albinism (OCA). The following information explains the study and which types of subjects are included and excluded from the study. Please contact their office directly with any questions.



Purpose: The purpose of this study is to evaluate and document physiologic and functional changes in visual performance and retinal function of patients diagnosed with albinism (a dopamine deficiency state) following a trial of oral Levodopa/Carbidopa treatment.

Detailed Description: In this study, the investigator proposes that the retina itself in albinism is deficient in dopamine, and vision improvement will occur as a result of improved retinal function in response to the deficient neurotransmitter dopamine. This study has a pre-test/post-test design in order to determine if improvement in vision is in response to replacement of deficiency (dopamine). The electroretinogram (ERG) testing and optical coherence tomography (OCT) will be critical determinants to confirm vision improvement as a result of improved retinal function, but are not primary outcome data. Main outcome measures will be collected at pre-treatment, 3 months, and 4 months. Change in visual acuity as measured in logMAR by Snellen or sweep visually evoked potential (SVEP) after 3 months of treatment is the primary outcome.

Subjects include OCA1a, OCA1b, OCA2, and unclassified OCA patients. OCA1a patients clinically are known to have the most impacted vision, and physiologically have the lowest (or absent) levels of tyrosinase function (Dopamine Production). All patients will be treated with Levodopa/Carbidopa 4 mg/kg/day in three divided doses.



It is important to note that subjects must have albinism (OCA), not ocular albinism (OA).

Further inclusion and exclusion information for the trial can be found on the Vision of Children Web site or you can contact the study coordinator directly.

Study coordinator: Angie Wealti
(608) 265-7557
or e-mail: wealti@ophth.wisc.edu

To access their Web page, you may link to it from the VOC Web site:
www.visionofchildren.org

...Vittoria, Continued from Page 1

When Andrea was asked to found and direct the Telethon Institute for Genetics and Medicine in Milan, we all moved back to Italy, and there I proceeded with studies aimed at understanding the nature and function of the protein product of OA1. In fact, typically the gene identification immediately gives you the possibility to perform precise diagnosis for a genetic disease, but therapy in most cases is still far away. To get to therapy, it is necessary to unravel several other steps, from genetics to cell biology, to developmental biology, to physiology etc, since you cannot correct what you do not know. Indeed, several teams of scientists typically specializing on different aspects are required.

My focus is the pathogenesis of Ocular Albinism at the cell biology level, which means understanding the function of the OA1 gene and protein in pigment cells, as an obligatory step required for any future attempt to establish the targets and evaluate the efficacy of pharmacological or gene therapy approaches. Indeed, understanding of the molecular mechanism by which loss of OA1 function leads to disease could be instrumental in identifying specific subcellular targets of pharmacological therapies. This possibility is particularly concrete given that OA1 is a G protein-coupled receptor and more than half of the drugs presently on the market (anti-histamine, anti-asthmatic or anti-arrhythmic drugs for instance) target this class of receptors or their pathways.

Given that the relevant molecular and functional consequences of OA1 deficiency in melanosome biogenesis are still unclear, it is necessary to precisely understand the role played by OA1 in order to evaluate any disease correction. In addition to this, during our studies, it came out that Ocular Albinism also represents an interesting model for understanding basic biological mechanisms, since the OA1 protein represents the first example so far of an exclusively intracellular G protein-coupled receptor. Therefore, by studying OA1, it is possible to withdraw more general implications for cell biology and molecular medicine, by enlightening signaling processes operating at other essential secretory organelles throughout the body. I think my main contributions to OA have actually been along this line, by understanding the receptor nature of OA1 and its involvement in melanosome motility, both for their relevance to OA and for cell biology in general.



As alpinism is a kind of "fight" with the Alps and with adverse natural elements, but especially a fascinating exploratory adventure, scientific research represents a never-ending fight for publications and funds, but also an extraordinary investigation of the "unknown." In real research, you cannot predict the results and should be prepared to find unexpected readouts. Even the knowledge obtained is not dogmatic, but remains a dynamic learning process, susceptible to corrections. So, despite previous know-how and plans, you need to have an exploratory spirit and be prepared to find new paths.

I first heard about VOC in 1995, just before identifying the OA1 gene, but it wasn't until 2000 when I started my own lab and that I obtained my first VOC grant. VOC was also critical during subsequent years, since thanks to the sensitization work that VOC did toward the NIH to promote grants dedicated to albinism, a call was announced by the National Eye Institute. I applied and finally obtained a R01 grant, which was fundamental for my work and career. Overall, VOC has always had a profound impact on my research and helped me significantly, particularly during "hard" times, which researchers have to face, when the availability of funds becomes restricted.

Unfortunately, compared to other diseases albinism is rare, so it is not extensively known and studied, and not many funds are available, particularly from the government. So it is critical that organizations like VOC are there to help patients and families, to communicate and sensitize the general public and the institutions, to support research either directly or indirectly, since without it we can't move ahead.

I also think that spreading of correct information is a critical issue and scientists should contribute to it. So during the last few years, I was particularly glad to participate as scientific advisor to an original photographic project on albinism, mainly conceived and realized by the photographer and journalist Silvia Amodio in collaboration with Elisa Tronconi, president of the Italian Association for Albinism (Albinit; <http://www.albinit.org>) (pictured above). With the purpose of enhancing the knowledge of the condition of persons with albinism by means of art, Silvia realized a series of intense and poetic portraits of these kids and adults with albinism, which were displayed by an itinerant exhibition and by a 2013 calendar. Considering the relevance of the final aim and the quality of the results, which already received several awards and positive appraisals, even from art critique, I am really happy I gave my small contribution to this battle against ignorance!

Helping to Improve Lives = Special Project

IMPROVING LIVES In line with the part of our mission that says we want to improve the lives of visually impaired individuals and their families, we have been providing low vision equipment to schools and individuals for several years. Through this "Project Vision Aid" program we've been providing schools and students with small handheld video magnifiers. The feedback from students/teachers is that they love them and it is convenient to be able to take it from class to class and home to do homework.

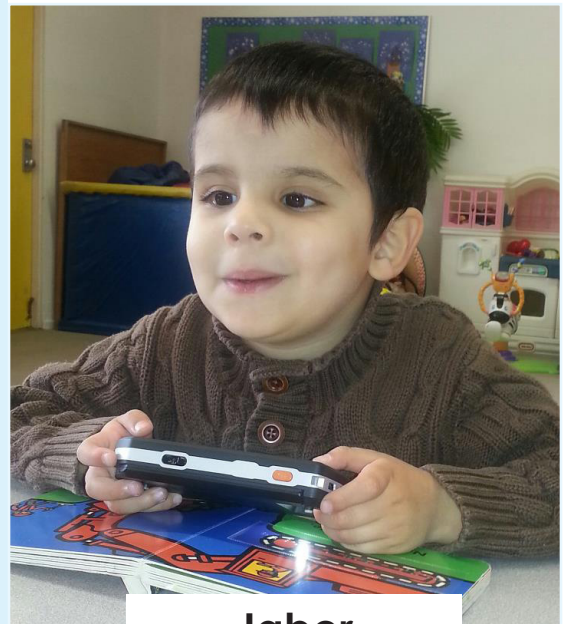
BLIND CHILDREN'S LEARNING CENTER This February VOC was able to present the Blind Children's Learning Center, in Santa Ana, CA, with 14 handheld magnifiers for their students thanks to a grant from the Allergan Foundation. The VOC staff was thrilled to be on campus to present three of them to young students at the school (Victoria and Jaber, pictured right were two of them). Having magnifiers at an early age will prepare them to be technology users throughout their lives. This has also created what we believe is a special relationship with the Center. Blind Children's Learning Center's President and Executive Director, Kathy Buehler, was ecstatic when informed they were selected as our partner and would be able to award their students the devices. "These magnifiers will help our low vision children in our preschool to identify letters and numbers. They will be instrumental to the children we serve in the school districts with their reading and math. We are extremely thankful to The Vision of Children for this donation," said Ms. Buehler.

AIDING ABILITY Children with vision impairments have the same potential as students with full sight. It's The Vision of Children's hope that having portable equipment at their fingertips will help to improve these students' lives. One San Diego middle-school teacher had this to say in response to the benefit her students are getting from the device: "This is awesome - it helps with the students' mobility and independence, which hasn't been possible with other technologies prior to this."

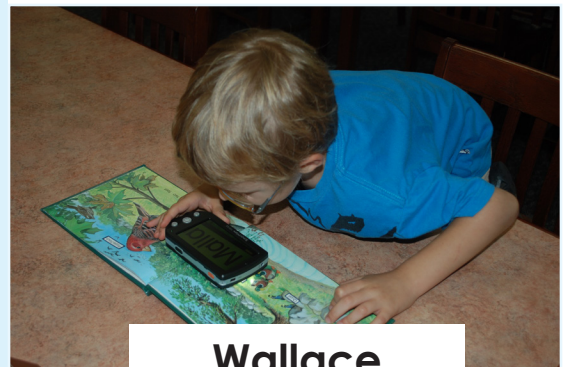
RECEIVE A MAGNIFIER FOR YOUR CHILD We received a grant recently which is enabling us to provide magnifiers to our Members, regardless of where they live. As mentioned in our fall newsletter and subsequent e-mail messages, we are now providing a Membership, which will help offset our administrative costs and enable us to put more dollars toward research. As a benefit of Membership, our paid members are eligible to apply for a free magnifier for their child. We will take applications through June and anticipate distributing them in August, which should be just in time for the new school year. Wallace, pictured right, attends Solana Vista Elementary and was awarded a magnifier in



Victoria



Jaber



Wallace

Spotlight On: Riley - Insights of Vision Loss

In our first *Insights of Vision Loss* spotlight video, created by her 17-year-old sister, Schade, we get to know Riley, a 14-year-old 8th grader at Correia Junior High in San Diego. She shares that her vision limits her ability to see the board at times in school, to play certain sports, and she gets questioned about whether she'll be able to drive—all familiar concerns to individuals and parents of individuals with low vision. She appreciates the work Vision of Children is doing to help her and others like her living with vision disorders and she is inspired by her Mom's recent fundraising efforts to help VOC (Cyndie is featured on p.3). Please check out the video on the VOC YouTube page to learn more!

<http://www.youtube.com/visionofchildren>



Chapman University Delta Gammas Shine Again With Holiday Fundraiser

In 2011 The Vision of Children and the Chapman University Delta Gamma collegians created an event, which is now a staple for the holiday season in Orange County. In early December, small pre-lit Christmas trees were distributed to individuals and various campus organizations at Chapman – each with the task of festively decorating them for auction. There were 30 beautiful and creative trees on auction, ranging from ornaments of pretzels (yes, pretzels!), to a Disney princess theme, to a Disney Cars theme, and lavishly decorated trees that sold for as much as \$200. The event was held at Legacy at Tustin Hills in the late afternoon and was complete with delicious desserts & beverages, an opportunity drawing, and the entertainment of an a cappella group from the university. It was attended by the collegians, their parents, their friends, local alumnae, and chapter advisors. Megan Powers, Director of Development & Communications, was also on-hand to let the attendees know about the work VOC is doing and how appreciative the Foundation is of their continued support (she too is a Delta Gamma).

Work is already being done to get trees donated for the 2013 event!

If this type of event is one you are willing to hold for VOC in your community, please reach out to us and we will give you the “how-to” for this easy, but very effective fundraiser.





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Become an "official" Member today - see why on pg. 5 & visionofchildren.org!

Jeans Day Sets New Record

An easy way for organizations to show their community spirit and provide a way for their employees to do something good, while FEELING good, is to hold a Jeans Day!

Ally Commercial Finance in New York City has been the perfect example of how it is done. They hold a Jeans Day for a different organization every month and November has been Vision of Children's month for several years now. This past fall they raised an unprecedented \$6,000 from their employees, which Ally then matched, providing \$12,000 to VOC in a snap!

Simply pick a day on which employees can make a donation to The Vision of Children Foundation in exchange for wearing jeans to work. Many companies incentivize employees to make additional donations by offering the opportunity to win prizes such as designer jeans. This encourages the employees to donate more and increases their chances of winning!

Employers who will then match what the employees contribute results in GOLD for just one day of employees getting the chance to show they care.



Tyler Schrag Update - Where Have the Years Gone?

In 2005 we published an article on Tyler, showing what a great entrepreneurial fundraiser he was... And he still is! He still has a business called "Sweet Tooth Candy," through which he raises funds for VOC.



The first check he sent to us was for \$20 and we recently received a letter and a check for \$100. He is still working and has also gained some inches, as you can see here! We see a bright future ahead. As Tyler says: "Goodbye. Work hard. God bless!" THANK YOU, Tyler - It's individuals like you who keep us going!