HOPE in FOCUS
Seeing a cure for blindness
Supporting the LCA and rare retinal disease community

# SEEING HOPE | Newsletter

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## Sofia Sees Hope Rebrands with an Eye to the Future

***By Rosanne Smyle***

Sofia Sees Hope this month unveiled a new name and logo — Hope in Focus — as the organization sets its course for the next decade of work to benefit the Leber congenital amaurosis (LCA) community.

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Embracing and encompassing, Hope in Focus is our new name: Embracing because research advancements fuel hope and ease feelings of isolation that often accompany a rare-disease diagnosis, and encompassing because our global advocacy reaches all of those living with LCA and other rare inherited retinal diseases (IRDs).

Our organizational namesake, Sofia, was a little girl with LCA in 2014. She has now come of age, and so have we. Throughout the last seven years, Sofia Sees Hope has transformed from a small NPO into a robust, international organization, and that's why we've taken the exciting step of evolving our name while keeping hope as its centerpiece.

"Hope is built on the bedrock of yearning; not an unrequited yearning, but the yearning for what we know *can be*," President and Co-Founder Laura Manfre said. "Hope is fuel, driving us to action. It binds us together and soothes our souls when darkness falls. Hope is our Polaris; the brightest star in the IRD constellation."

Hope also is empowering and a strong motivator. It is the role of Hope in Focus to make sure we hear those voices of the patients, families, and community, and we help craft compassionate, forward-thinking policy.

"Without hope there is not a chance you're going to advance treatment," Manfre said. "We cannot sit back and wait for it like manna from heaven because that's just not going to work."

### WHY CHANGE THE NAME?

As Sofia Sees Hope grew from its beginnings, so did its reach geographically and genetically with those living with LCA and

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## *From the Founder*

*Sofia Priebe & Laura Manfre*

When we started Sofia Sees Hope in 2014, we named the organization in honor of our daughter, Sofia, who was born with Leber congenital amaurosis. The truth is, though, we've been working on behalf of *all* the Sofias in the world: Children and adults living with LCA and other rare inherited retinal diseases.

You know us as an organization that adeptly moves between advocacy and science, keeping those we serve **in focus** each step of the way. This is our way of bridging the unnecessary chasm between the people who inform the science, and the science that transforms the people. **Hope** is born of connecting these two sides of the same story. In that spirit, please let us introduce you to

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## From the Founder

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the new name of our advocacy organization: Hope in Focus.

Our mission remains unchanged: We continue to generate awareness, raise funds for research, and provide outreach, support, and education to those affected by rare retinal disease. Fostering hope remains central to what we do.

Honestly, when we first started, all we had was hope; there weren't any treatments on the horizon that we could see. But in just three years, the first treatment made it to market.

We witnessed that development process and now we've got a half-dozen treatments for LCA ready to follow on the heels of that science. And it's not just about the science anymore: It's about the biotech industry and the drug development process; it's about patient community involvement and policy and manufacturing, and all these other things we're learning about and we're going to keep learning about in our goal to drive research to find treatment and cures for these rare diseases.

When we think about hope and our mission, I am reminded of Elie Wiesel's words on the subject: *"Hope is like peace. It is not a gift from God. It is only a gift we can give one another."*

When you or a loved one receives a rare disease diagnosis, it can feel like the floor has fallen out from under you. There may not be any good answers to your questions, but there is hope that will change. That hope is a gift we need to give. Hope is a powerful motivator. It lifts us up and moves us to action. Those actions are turning our hopes, my hope for my child, into a reality.

We are proud of our past, we are optimistic about our future, and we are ever thankful to share this meaningful work with every one of you as we move forward with Hope in Focus.

With gratitude and focus,

## Sofia Sees Hope Rebrands with an Eye to the Future

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IRDs. The development and 2017 federal approval of LUXTURNA®, a vision-restoring gene therapy, marked an incredible milestone in the rare retinal disease world.

Several factors powered the name change. Sofia Sees Hope established steady grant funding and strong corporate partnerships over the years to support advocacy programs, such as our LCA Family Conferences, quarterly newsletter, and monthly webinars.

"We are grateful to our grantors and corporate partners, and, for the most part, that funding has remained steady and that's what we use for outreach and for continuing to build a sustainable organization," Manfre said.

Our advocacy group forged vital relationships with global organizations like Foundation Fighting Blindness and Retina International and with pharmaceutical and biotechnology companies, resulting in remarkable research advancements. These include improved access to genetic testing and the development of more than 30 clinical trials into retinal disease research.

"We've reached this level of organizational maturity where we're well-known enough that we can manage a name change and continue to provide even better service to the community," Manfre said. "We are well-positioned for the future and believe this name change will only help us to be more successful."

### UNINTENTIONAL OPPORTUNITY OF TIME

Manfre said she didn't necessarily want her daughter's name attached to the organization because from the beginning it was about so much more than her. After Sofia received her confirmed genetic diagnosis of the gene causing her vision loss — IQCB1 — her family was asked to fundraise.

"Every year I would step back and question it. Is it time to change our name? It was never about Sofia specifically," she said. "Since our incoporation, we provided funding to support a variety of research initiatives, including My Retina Tracker® Program."

But then the COVID-19 pandemic set in, giving an unintentional opportunity to press the pause button for time to re-evaluate.

"COVID gave us time, although that's not how we would have wanted to do it."

The name Sofia Sees Hope served our organization well once we established we were reaching out to people like Manfre's daughter, Sofia, and people living with any one of the 27 known forms of LCA and a host of IRDs.

Now in the eighth year since our founding, research has advanced exponentially, and we wanted to be clear that we are there for the whole LCA and IRD community" she said.

"We are not changing who we are or what we do. We're just changing the name."

Manfre said the name change also relieves the pressure on Sofia that comes with having her name be part of the organization. Sofia, having her name be part of the organization, which she has supported at fundraisers, educational events, and most recently by leading a book club for middle schoolers with LCA and other visual impairments.

## THE PROCESS BEHIND OUR NEW NAME

We had amazing help with finding the right name.

Bonnie Southcott led the three-month project. Southcott is Director of Patient Engagement at Toolhouse, a digital marketing firm in the life sciences sector, based in Washington state.

Under her guidance, we developed a new name, Hope in Focus, a new tagline, "Seeing a cure for blindness," and a new logo.

"I think it's important to know that the organization itself hasn't changed. The (new) name is more reflective of the greatest audience that they serve and of their vision for the future," she said.

"The other piece of it is that it takes away one of the questions, and that question was 'Who's Sofia?'"

Sofia represented people with LCA, but some might not get that and think twice before reaching out to our organization, thinking, 'I don't know who Sofia is, the organization might not be right for me.'

"That only has to happen once to have an impact," Southcott said. "To take away that question was key."

She describes the essence of rebranding or renaming as carefully identifying a brand's DNA and then capturing it in the new words, look, and feel of the name, the logo, and the tagline. By involving representatives from each

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# Sofia Sees Hope Rebrands with an Eye to the Future

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stakeholder group and planning a careful rollout, she said, the organization's supporters, constituents, and staff become champions of the reimagined brand.

"Hope: That was almost like a life raft that people jumped into. You have to focus on hope. It is too central for what this organization stands for."

The name also reflects the human connection the organization makes with the LCA and IRD communities: "The warm embrace that Hope in Focus stands for."

The name also differentiated our group nicely from others in the field and that's important for messaging, important for fundraising, and important for growth, she said.

"Some do offer that sense of hope, but you don't get that from the name. The new name really had to convey that, and it had to underscore that the purpose is not only Sofia, but all the Sofias.

"The hope component — that sets us apart."

Science and research comprised the other piece folding into the mix, thinking about microscopes and getting a clearer focus on treatments and cures, especially with one gene therapy on the market and more developing research in clinical trials.

Southcott and her colleague Chance Martenson began with 40 possibilities for names and narrowed them to 12 after conversations with our founders, board members, staff, donors, and the people we connect with in the LCA and IRD communities. From there, they recommended three for consideration.

The name change or rebrand of our organization stood out among other projects she has done.

"It was more an evolution of what existed, as opposed to a revolution or the creation of something brand new, where nothing existed before."

Finding the right name also meant not disenfranchising people involved with our advocacy group.

"There is this real sense of dedication to the organization and a yearning to protect it from anything else, Southcott said. At the end of the day, it had to serve the people that support it and use it."

## OUR FUTURE

We at Hope in Focus will expand the reach in our advocacy efforts and continue to grow as a small team doing big things, Manfre said. The vision for Hope in Focus is far reaching, and we are developing exciting ways to engage our community members and bring them together as we support them, and they support one another.

"We've been powered by a small team of part-time consultants and volunteers, and we've just recently brought on board a full-time development and outreach director," Manfre said. "To be able to continue to expand to meet the needs and the demands of the LCA and IRD communities, we need to keep growing. I view this as the first step in that growth.

"We're checking all the boxes, working to fill unmet needs. This is much bigger than me and much bigger than Sofia. We only began something that is going to continue to grow, and we're very excited about where it will go."

## We need your support.

**Help us continue our work to advance treatments for LCA.**

Visit www.hopeinfocus.org, or send a check to Hope in Focus P.O. Box 705, Ledyard, CT 06339

HOPE in FOCUS

Seeing a cure for blindness

# Encouraging Early Report for Three Patients in LCA1-GUCY2D Gene Therapy Clinical Trial

*The Retinal Degeneration Fund of the Foundation Fighting Blindness is funding development of the emerging gene therapy*

***By Ben Shaberman***
***Senior Director, Scientific Outreach & Community Engagement***

FOUNDATION FIGHTING BLINDNESS

A clinical research team led by Samuel Jacobson, MD, PhD, at the University of Pennsylvania, observed vision improvements for the first three patients with Leber congenital amaurosis 1 (LCA1) treated with an emerging GUCY2D gene therapy in a Phase 1/2 clinical trial. The treatment also demonstrated a favorable safety profile.

Atsena Therapeutics, a clinical-stage gene therapy company in Durham, North Carolina, is developing the gene therapy. The journal *iScience* published online the early results for the trial.

The first three patients receiving subretinal injection doses in the trial were adults with advanced vision loss but with some remaining retinal structure. All received the lowest dose of the treatment.

Patients 1 and 2 had improved rod function as measured by full-field sensitivity, a test appropriate for those with advanced loss because it doesn't require the patient to fixate. Rod photoreceptors provide peripheral vision and vision in dim settings. The visual acuity of Patient 3 improved from 20/400 to 20/200, indicating improved cone function.

The therapy was created in the laboratory of Atsena Founder and Chief Scientific Officer Shannon Boye, PhD, and her husband, Founder and Chief Technology Officer Sanford Boye, MSc, at the University of Florida.

Atsena is being funded by an investment from the Foundation's Retinal Degeneration Fund, a venture philanthropy fund for emerging treatments in, or moving toward, early-stage clinical trials.

"We are encouraged by these early results for Atsena's LCA1-GUCY2D gene therapy and look forward to additional results for patients with better vision who will receive higher doses," said Benjamin Yerxa, PhD, Chief Executive Officer at the Foundation. "Gene therapy is a good potential approach for many GUCY2D patients because they have remaining retinal structure despite advanced vision loss."

LCA is the most common cause of blindness in children, impacting two to three per 100,000. LCA1 is caused by mutations in the GUCY2D gene and results in early and severe vision impairment or blindness. LCA1-GUCY2D is one of the most common forms of LCA, affecting roughly 20 percent of patients who live with this inherited retinal disease.

*Visit **FightBlindness.org** to stay abreast of the latest research advances for LCA and other IRDs.*

# Dating With Vision Loss: Be Proactive, and Practice

*Jack and his guide dog, Baloo*

Of all the writing I've done for this newsletter, writing this column has been the most challenging. I wish I could tell you that I've figured out how to overcome all the challenges of dating with a visual impairment, but the truth is I haven't.

Some of these challenges are self-imposed. I've figured out how to overcome these. To explain, I used to worry that no one would want to date me because of my visual impairment. In the past, I missed out because of these fears and that is something I regret. Thankfully, I no longer worry about this. The best advice I can give someone who has similar fears is to try. By this I mean ask out your crush and don't be afraid to ask for someone's number. It is scary to do, but doing these things is scary for everyone and the best way to make them less scary is through practice.

My second girlfriend taught me a valuable lesson about communication. About a month into our relationship, she asked me, "Beyond saying 'Can I kiss you, Jack,' how do I communicate that I want to kiss you?" Clearly there were some visual cues that I was missing. So, we learned to communicate by touch.

As much as I appreciated that she thought to ask this, I've learned that it is better to proactively discuss the challenges that come with a visual impairment and how I overcome them. It is likely that your date hasn't been with someone with a visual impairment before, so by speaking openly you'll help them feel comfortable and, more importantly, provide them with an open door to ask questions.

The other benefit of doing this is that the questions your date asks about your visual impairment are a good judge of character. From my experience, dates who don't ask me anything about my visual impairment when I bring it up are either not very thoughtful or not interested in the date. On the other hand, dates who spend the rest of the night asking me every question under the sun about my eyes don't tend to see past the visual impairment. The sweet spot seems to be somewhere in the middle.

On the topic of disclosure, I thought I'd wrap things up by sharing my thoughts on when you should let your date know that you have a visual impairment if they don't already know you. My rule of thumb is that my date should know before our first date, but it shouldn't be the first thing that I tell her about myself. I don't want it to be the first thing I share because my vision loss doesn't define me, and I share before a first date so that my date isn't surprised.

Good luck on your next date!

*Jack McCormick graduated in 2018 from Canada's Wilfrid Laurier University in Waterloo, Ontario. He was diagnosed in high school with LCA2. Jack is a Hope in Focus ambassador, helping people living with LCAs and IRDs.*

*You can read his blog at...*
***jackdamccormick.wordpress.com***

# COVID Pivot Results in Successful 2021 Webinar Series

We launched a web series this year called "**L**et's **C**hat **A**bout..." and we did just that, chatting with various experts about gene therapies, federal drug approval requirements, genetic testing, natural history studies, patient registries, self-advocacy, and the patient voice.

We covered a lot of ground on Leber congenital amaurosis (LCA) and other rare inherited retinal diseases (IRDs), and we will cover even more in the future, including CRISPR's gene-editing technology and ProQR's work in treatments for IRDs.

We've developed the series with those living with LCA and IRDs in mind, and we invite all members of our community, including those in research, industry, and the regulatory communities to join any of the sessions, as we look ahead to a common goal of advancing treatments for rare retinal disease.

Ben Shaberman of the Foundation Fighting Blindness kicked off the series, taking his audience from identifying the RPE65 gene in 1993 to the 2017 federal approval of vision-restoring gene therapy for that form of LCA.

Dr. Wiley Chambers of the Food and Drug Administration discussed the urgency of creating a vaccine against COVID and said the development of vision-improving gene therapies also meets fast-track requirements because of the seriousness of vision loss.

Geneticist Emily Place of Massachusetts Eye and Ear talked about her role as a genetic counselor, navigating the complex world of gene mutations, genetic testing, and genetic diagnoses. She described creating relationships with patients as a true privilege.

Jonathan Stokes of AbbVie brought to light the patient perspective, saying, "For me, the question is always the 'So what? What does that mean to the patient?'" He develops and evaluates clinical outcomes of assessment used in clinical trials to substantiate the benefits of treatment.

Todd Durham of the Foundation Fighting Blindness discussed the importance of patients joining the My Retina Tracker® registry and helping science by driving research to improve quality of life and find treatments and cures.

Tami Morehouse and Jack McCormick shared heartfelt stories of courage as they learned to ask for help in life. Tami had to dive into self-advocacy at age 55 to find a new job, while 24-year-old Jack hid his vision loss, until he got his beloved guide dog.

To watch the series and read the stories, visit our website at hopeinfocus.org, and choose "Let's Chat About" under the For Families drop down.

# Events

**DO YOU HAVE AN EVENT YOU WANT TO SHARE? LET US KNOW!**
**Email rosanne@hopeinfocus.org with the information and a link.**

### VisionWalks
**Oct 24, 2021 • San Diego**
**Oct 30, 2021 • Boston**
**Nov 6, 2021 • Los Angeles**
**Fightingblindness.org/events**

Foundation Fighting Blindness's VisionWalk has raised more than $58 million to fund sight-saving research.

### 2021 Bidding in the Dark
**Oct 22–29, 2021 • Virtual • hopeinfocus.org**

This virtual event is critical to the work of our organization and our ability to raise funds for treatments for rare inherited retinal disease (IRD), while providing a tiny snapshot of what it might be like to live with a rare IRD. Step outside your comfort zone and connect with our mission by bidding on items you cannot see.

### WEBINAR: Let's Chat About… ProQR's work in treatments for inherited retinal disease
**Nov 9, 2021 • 1–2 p.m. ET • hopeinfocus.org/event**

Join us as we chat with Daniel de Boer, Founder and Chief Executive Officer of ProQR. Daniel is a passionate advocate for rare disease patients. After one of his children was diagnosed with a rare disease, he started ProQR to develop RNA therapies for rare diseases.

### 2021 RARE Health Equity Summit
**Nov 17–19, 2021 • Philadelphia, PA**
**globalgenes.org/event/rare-health-equity-summit/**

This event connects stakeholders from the rare disease community to find ways to deal with inequities in the care of patients with rare diseases, specifically focused on shortening the diagnostic odyssey, reducing racial disparities in care, and building more inclusive research programs.

### Giving Tuesday
**Nov 30, 2021 • givingtuesday.org**

Every year on the Tuesday after Thanksgiving, people take the time to kick off the holiday season by giving back to their community. Stay tuned for our Giving Tuesday message.

### WEBINAR: Let's Chat About… MeiraGTX's work in IRDs and what's new in clinical trials.
**Dec 8, 2021 • 1–2 p.m. ET • hopeinfocus.org/event**

Join us to chat with Michel Michaelides, MD, a founding member of MeiraGTx and Professor of Ophthalmology, UCL Institute of Ophthalmology in Dept. of Genetics. He is the Principal Investigator of 3 active interventional clinical trials and has 10 on-going ethically approved studies.


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**To learn more about Hope in Focus, visit www.hopeinfocus.org.**

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