

# SEEING HOPE | Newsletter

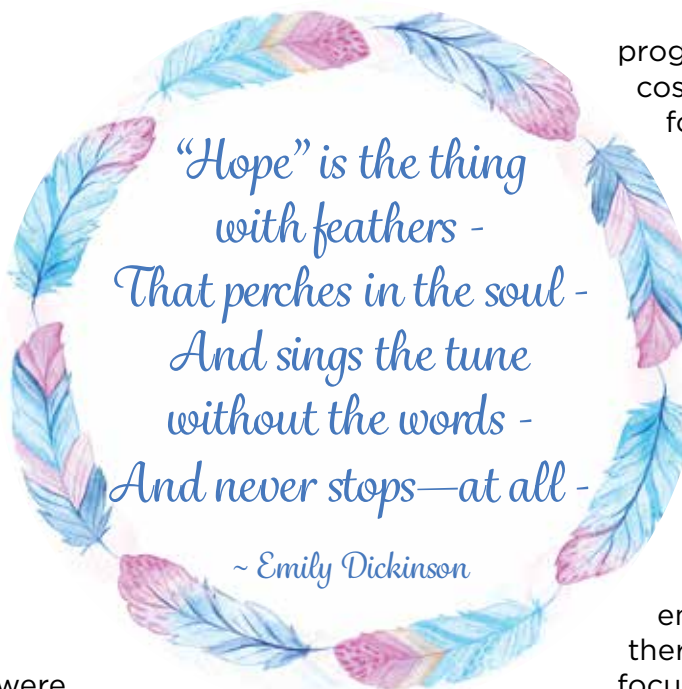
Years ago, when we first received our daughter Sofia's diagnosis of Leber congenital amaurosis, a rare inherited retinal disease that causes low vision and blindness from birth, we weren't given much hope. At that time, there were no approved treatments or trials underway, and we didn't know of anyone else with the disease.

In fact, we were told the best we could hope for was that the disease would progress slowly, and if she were lucky she would retain some perception of light through the end of high school, maybe even into her twenties.

Sitting at our kitchen table with close friends, we decided no hope wasn't an option.

Back when we were on our journey toward a diagnosis, it felt like it was all uphill. It was hard to get genetic testing. It was hard to get a genetic diagnosis. It was hard to find doctors well-versed in Leber congenital amaurosis (LCA). It was hard to find other parents and other children with the same disease.

That's why we started Sofia Sees Hope in 2014. And since then, we have grown a community of support, advocacy and education.



We have funded research around the globe that has resulted in clinical trials for treatments. We have paved the way for many families to receive genetic testing and counseling, because as important as we know hope is, we know knowledge is power.

Sofia Sees Hope supports innovative, early-stage proof of concept research programs and inherited retinal disease (IRD) genetic testing through fundraising events and individual donations. We partner with the Foundation Fighting Blindness (FFB) to designate our research contributions, and to ensure that free genetic testing and counseling continues to be available to individuals through FFB's My Retina Tracker® program. The

program is an open access, no-cost genetic testing program for individuals living in the United States with a clinical diagnosis of an IRD.


To date we have provided more than \$383,000 for research and testing. And we've done it all because of you.

Fifteen years ago, there were only one or two clinical trials underway for emerging therapies. Today, there are dozens of companies focused on IRDs and more than 40 ongoing clinical trials for potential IRD treatments.

So this issue of our newsletter is dedicated to just that: the concept of hope. You will find a donation envelope tucked inside. In a year that has challenged all of us perhaps more than we ever have been, we hope you can help us to continue our mission. Hope, after all, never stops.



*Sofia Priebe & Laura Manfre*

  
Laura

# Sofia Sees Hope Ambassadors Deliver

*By Rosanne Smyle*

We are fortunate to have an amazing network of passionate supporters who enhance our mission in their communities across the globe in their work as Sofia Sees Hope Ambassadors.

Our Ambassadors give encouragement to Leber congenital amaurosis (LCA) patients and caregivers, they attend conferences, keep up with research, and share their stories and experiences to help others.

These emissaries send love, hope and support to families as they begin their journeys of living with LCA or other inherited retinal diseases (IRDs) and into the future.

“Our Ambassadors are caretakers, patients, clinicians and friends from across the United States, in Canada, Mexico and the Netherlands,” said Ashley Luppold, Ambassador Program director. “Each team member brings their own unique experience and talents to the team.

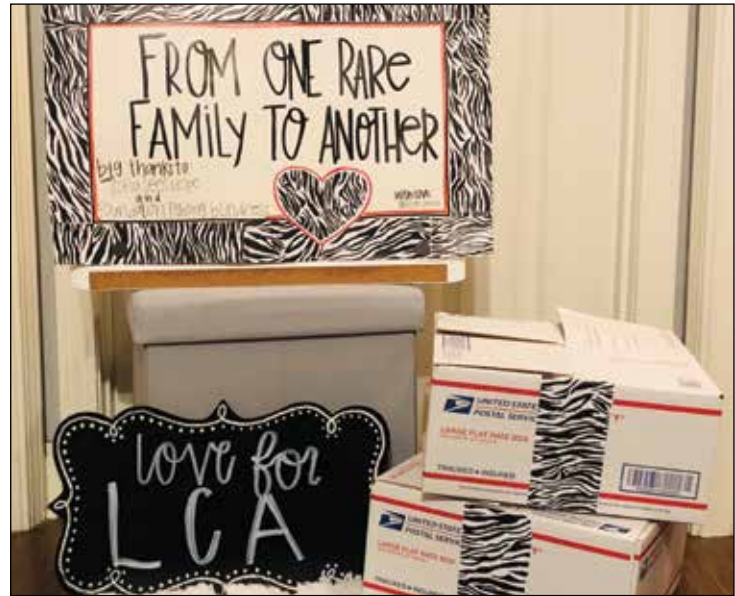
“This program simply allows us to tap into all that energy, and keep everyone informed and aligned around new research, resources and initiatives,” she said.

The Ambassador Program encompasses four focus areas that correlate with our mission—Family Outreach and Support, Education and Research, Fundraising and Development, and Advocacy.

“We ask our Ambassadors to find ways in which they feel they can contribute across one or more of those areas,” Luppold said. “We provide the team with resources to support their work in each of our focus areas, and the rest is up to them.”

Here’s a look at some of the many contributions and connections made by our Ambassadors:

Ambassador **Ashlyn Lincoln** took her experience of creating and sending care packages to her husband while he was deployed with the Marines to another level



*Ashlyn care packages*



*Ashlyn package contents sampling*



*Care Package Photos of Gunner with letter to families*



by fashioning personalized care packages for families living with LCA or IRDs.

“I am a care-package professional!” she said.

Living in a suburb of Nashville, Tennessee, Ashlyn and her husband, Axel, are parents to 6-year-old Gunner, who was born without vision, and 8-year-old Ace, who is sighted. Doctors diagnosed Gunner at 6 months with LCA10 caused by a mutation in his CEP290 gene.

“Since becoming an Ambassador with Sofia Sees Hope, I have prepared and slightly decorated the boxes families will receive,” Ashlyn said. “After seeing how many visually impaired-friendly books, toys and games that Gunner has outgrown, I felt it was time to pass these things on to other families, just as they were passed down to me.”

She includes books, games, tactile paper for fine motor skills, along with photos of Gunner as he has grown and a message to families that reads in part:

“It seems just yesterday my journey began as having a child with a Visual Impairment. I was overwhelmed, exhausted, emotional, determined and lost. All at once.”

Ashlyn goes on to explain that at her first event for families living with vision loss, she received love, support and a helping hand. They helped entertain Ace while she tended to an overwhelmed and overstimulated Gunner. They passed on books and games and toys and gave her information for more resources.

“Let me be that support for you today,” she tells families. “Please, take it all! I am passing on with love, just as they were passed onto me..”

.....

Ambassador **Scott Soady** lives in San Diego, Calif., with his wife, Heather, and daughters Gillian, 8, and Juliet, 5. Juliet has LCA7, caused by a mutation in her CRX gene. While LCA is a rare disease, LCA7 is even more rare.

Being on the West Coast, Scott has attended conferences there on behalf of Sofia Sees Hope, headquartered on the East Coast.

He represented our organization at two Global Genes RARE Patient Advocacy Summits in California.

“This is a conference where the only common denominator is that patients and families are dealing with rare genetic disorders with an occurrence of less than 100,000 in the United States,” he said. “These are diseases that no one knows about, just as no one heard of LCA before they received a diagnosis. *Continued on page 4*”



*Heather Soady holds Juliet as her husband, Scott, daughter Gillian and friend Jessie take part in a Foundation Fighting Blindness VisionWalk in San Diego*



*Scott Soady with Gillian and Juliet*



*Juliet Soady*

# Sofia Sees Hope Ambassadors Deliver

Continued from page 3

“Even with the diversity of disorders, there are themes that are common that we (in the LCA and IRD realm) would recognize: difficulty obtaining a diagnosis, knowing more about the disorder than doctors, the lack of any treatment for the disease, the isolation you feel when initially diagnosed, and trying to find resources and a community.”

Scott has also taken part in Sofia Sees Hope’s Family Connections programs.

“It is always rewarding to talk to families that are newly diagnosed,” Scott said. “I will be forever grateful to have gotten support from (Sofia Sees Hope founder and co-president) Laura (Manfre) and others when Juliet was first diagnosed.”

Ambassador **Jack McCormick** of Ontario, Canada, graduated from Wilfrid Laurier University in Waterloo, Ontario, in 2018. Diagnosed with LCA2 due to a mutation in his RPE65 gene, Jack is a passionate advocate for inclusion and accessibility on all fronts. He initially tried to hide his blindness but that all ended when he got Jake, his beloved guide dog. Jake immediately made Jack feel like he was wearing a sticker that said, “Hey, I can’t see,” which led to acceptance. And acceptance led to respect and opening pathways to social accessibility, that, in turn, helps fuel research.



Jack McCormick

Jack encourages people with blindness to tell their stories.

As an Ambassador, Jack tells *his* story in every “Seeing Hope” newsletter, covering topics ranging from technology that helps the visually impaired to navigating the pandemic.

From the Cleveland area, Ambassador **Tami Morehouse** made research history when at age 44 she participated in a clinical trial for gene therapy for LCA2 (RPE65). Doctors injected under her retinas a human-engineered virus that gave instructions on how to produce an essential protein for vision. Spark Therapeutics developed the drug that was marketed as LUXTURNA® following Federal Drug Administration approval in December 2017.



Tami and Mike Morehouse

Tami and **her husband, Mike**, who also is an Ambassador, attended the Foundation Fighting Blindness VISIONS 2018 conference in San Diego and worked in the Sofia Sees Hope booth, sharing information on the clinical trial process, LUXTURNA®, the My Retina Tracker® genetic registry administered by the Foundation, and resources offered by Sofia Sees Hope.

She served as a panelist at our LCA Family Conferences in 2018 in Mystic, Connecticut, and 2019 in Philadelphia and privately talked with LCA patients and their families about frustrations with a lack of diagnosis, daily living, fears surrounding clinical trials and her experiences with them. She has attended our Dinner in the Dark gala as well.

“During the dinner, I was approached by two different mothers, who wanted to share that their young children had LCA/RPE65, had been treated with LUXTURNA® and were now going to be able to see,” she said. “On each occasion, the moms broke down in tears as they were telling me that their formerly visually impaired child can see because of my participation in the trial. It was a very rewarding but humbling experience that I still treasure.”

Sofia Sees Hope thanks all of our Ambassadors for giving the gift of hope.



# Reflecting on the Trajectory of IRD Research



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

**FOUNDATION FIGHTING BLINDNESS**

When I joined the Foundation Fighting Blindness as a science writer in 2004, I really didn't know what I was getting into. I knew nothing about the retina, let alone the complex and diverse world of rare inherited retinal diseases (IRDs) that includes Leber congenital amaurosis (LCA). But the research for treatments was cutting-edge and compelling, so I was excited to dive in and learn.

My early assignments were writing about laboratory studies coming out of academic labs. There were virtually no companies in the IRD space and only one or two clinical trials underway for emerging therapies. But there were a lot of studies of genetically engineered mice and rat models of IRDs for gaining a better understanding of disease pathways and testing potential treatments.

Truth be told, I often wondered if and when rodent-tested therapies were *really* going to make it into human studies and out to the people losing vision. But the scientists conducting the research were mind-blowingly smart and innovative, so I figured they knew what the heck they were

doing. With a master's degree in poetry, who was I to judge?

Fast forward about four years: I was in my hotel room in Fort Lauderdale—there for the annual Association for Research in Vision and Ophthalmology conference—when my manager called and told me three research groups just reported vision improvements in young adults treated with RPE65 gene therapies in Phase 1/2 clinical trials. That was the breakthrough we'd all been waiting for.

People, rather than animals, with severe vision loss were now seeing significantly better. It was the first time an IRD treatment had worked in humans. I will never forget the headline for the article I immediately wrote: "Now They See." (Note: One of those RPE65 gene therapies later became LUXTURN<sup>®</sup>, the first FDA-approved treatment for the eye or an inherited condition.)

After many years of painstaking work, our hope for treatments and cures had finally begun evolving into promise.

There have been several other aha! moments in the ensuing years, but I distinctly recall cathartic encounters at the 2019 American Society of Retinal Specialists in Chicago. As I perused the snack table during breaks (the accomplished snacking professional that I am), several representatives from biotechs developing IRD therapies—companies I'd never even heard of—came

up to introduce themselves to me and tell me about their emerging IRD treatments. They didn't know me or my role, nor had I previously known them; they were just eager to connect with someone from the Foundation Fighting Blindness to get on our radar screen.

I realized then I couldn't keep track of all the companies (dozens) focused on IRDs and clinical trials (40-plus) underway for potential IRD treatments. But being overwhelmed felt incredibly good, and it meant more good news likely was on the horizon for saving and restoring vision.

While mouse studies are as critical as ever, I can't remember the last time I wrote an article about one. That's because most of my writing is now dedicated to reporting on advances, including encouraging vision improvements, being made in human studies.

Make no mistake: Much more work needs to be done before we eradicate the myriad IRDs affecting millions of people across the globe. And, of course, we cannot get more therapies across the finish line fast enough. But when I look at how incredibly far we've come since those early days of mice and rats, I have no doubt we are well on our way to breaking many more ribbons soon.

Visit [FightBlindness.org](https://www.fightblindness.org) to stay abreast of the latest research advances for LCA and other IRDs.

## HOPE IS BELIEVING THE FUTURE CAN BE BETTER

I must admit, when I learned that my vision was slowly deteriorating, I lost hope. I didn't know what the future would bring. I knew that with worse vision came more accessibility barriers and was worried that these barriers would prevent me from achieving my dreams.

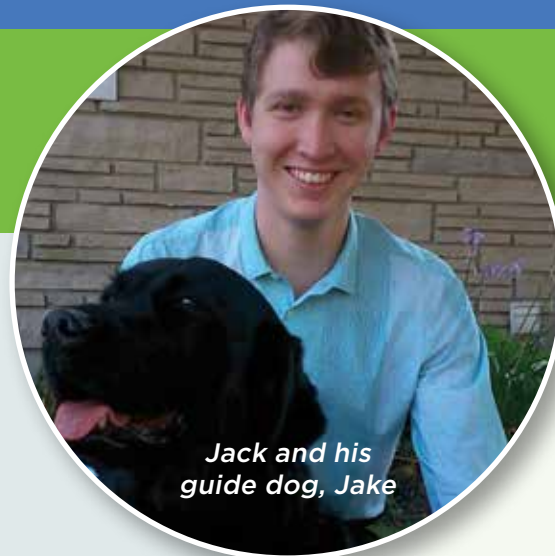
But my hope soon returned. I knew that I needed to make a choice. I could either feel bad about myself or do something to make a difference. I chose the second and began to plan. For me, hope is about believing the future can be better.

I wasn't sure if there was anything I could do about my vision. Genetics had made up its mind. The science was close to a treatment, but I couldn't count on it. That being said, many treatments were near enough to becoming reality that they would definitely affect many. This was my opportunity. I began raising money for sight-saving research, speaking at events and advocating for treatments. I've learned that snails move faster than medical research. It is frustrating and easy to lose hope. But, a snail's pace is still progress!

For example, LUXTURNA®. The first FDA-approved gene therapy for an inherited retinal disease was approved in the US long before it was approved here in Canada. I'm pleased to say that it was finally approved by Health Canada in October 2020. I have the type of LCA that LUXTURNA® treats, and if the provincial government agrees to fund it, I could receive this treatment soon.

The disease that causes my vision loss is advanced. LUXTURNA® will come nowhere close to giving me perfect vision. I've learned that having hope for restored vision and hope for life after vision loss are equally important. The best parts of life are not lost with vision loss. I have an amazing life. I have a lot to be thankful for—a good job, an incredible family, fantastic friends and opportunities to participate in accessible activities like sailing and skiing. I hope that you continue to have hope for life after vision loss too.

*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 Sofia Sees Hope ambassador, helping people living with LCAs and IRDs. You can read his blog at [jackdamccormick.wordpress.com](http://jackdamccormick.wordpress.com)*



*Jack and his guide dog, Jake*



*Jack and his family*



# EVENT CALENDAR – WINTER 2020/2021

## **Giving Tuesday December 3, 2020 Global**

Giving Tuesday is an international event dedicated to charitable giving. Organizations around the globe, including Sofia Sees Hope, raise funds to support their missions.

## **Foundation Fighting Blindness / Music to Our Eyes**

**December 10, 2020  
Live-streamed Event**  
[fightingblindness.org/  
events/music-to-our-eyes](https://fightingblindness.org/events/music-to-our-eyes)

The Foundation Fighting Blindness presents Music to Our Eyes, part of a live-streamed series of conversations and concerts in which artists discuss their experiences with vision loss and their commitment to creating music that celebrates diversity and belonging. The series is presented in partnership with Two Blind Brothers, founded by Bradford and Bryan Manning, who started the apparel company to create ultra-soft clothing, build community, and donate 100 percent of profits toward research curing blindness.

## **Rare Disease Day February 28, 2021**

Rare Disease Day takes place on the last day of February each year. The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.

## **National Organization for Rare Disorders (NORD) 2021 Rare Disease Day Virtual Advocacy Events**

**February 28, 2021  
Virtual Events • [rarediseases.org/rare-disease-day](https://rarediseases.org/rare-disease-day)**

Rare Disease Day takes place on the last day of February each year, and every four years, it ends up on the rarest day of the year! Rare Action Network volunteers are planning their 2021 Rare Disease Day Virtual Advocacy Events to raise awareness with the public and decision-makers about rare diseases and their impact on patients' lives. All events occur in February and March and will be virtual because of COVID-19 pandemic concerns. Check for details in January.

## **Foundation Fighting Blindness / Hope from Home: A United Night to Save Sight**

**February 28, 2021  
Virtual Event • [fightingblindness.org/events](https://fightingblindness.org/events)**

Hope from Home is an exciting and interactive virtual experience celebrating Rare Disease Day and featuring incredible entertainment, amazing auction items, and the opportunity to connect with others from the comfort of your own home. More details in February.

## **Foundation Fighting Blindness Investing in Cures Summit March 2021 • [fightingblindness.org/events](https://fightingblindness.org/events)**

The Foundation and its venture philanthropy investment arm, the Retinal Degeneration Fund, are in

the early planning stages for their Investing in Cures Summit. The event features presentations by the world's leading industry, clinical and retinal science innovators.

## **National Organization for Rare Disorders (NORD) Living Rare, Living Stronger Patient and Family Forum**

**June 2021 • [rarediseases.org/  
living-rare-forum](https://rarediseases.org/living-rare-forum)**

Living Rare, Living Stronger is an annual patient-focused conference, providing patients and families with practical tools for living their best lives with rare diseases and giving physicians and medical students insight to inform their practices and studies. Check details in January.

## **Global Genes Rare Drug Development Symposium**

**June 2021 • [globalgenes.org/  
event/rare-disease-drug-  
development](https://globalgenes.org/event/rare-disease-drug-development)**

This two-day interactive symposium, in partnership with the Penn Medicine Orphan Disease Center, focuses on educating beginners and advanced participants on the drug development process. Members of the rare disease community are encouraged to explore their roles in the rare disease drug development process in new and innovative ways. This personalized experience is dedicated to empowering the rare disease community to continue to push forward on necessary treatments and cures, despite experiencing setbacks because of fast-tracked research into COVID-19 treatments.

**SPECIAL EDITION**



**THIS NEWSLETTER IS MADE POSSIBLE BY THE GENEROSITY OF:**

- Spark Therapeutics
- MeiraGTx
- Janssen Pharmaceutical Companies of Johnson & Johnson

**Seeing Hope Newsletter Staff**

- Elissa Bass, Editor
- Rosanne Smyle, Writer
- Gina Morin, Graphic Designer

To learn more about Sofia Sees Hope, visit our website at [www.sofiaseeshope.org](http://www.sofiaseeshope.org).

The *Seeing Hope* newsletter is published quarterly by Sofia Sees Hope, a 501(c)(3) patient advocacy organization dedicated to generating awareness, raising funds for research, and providing education and outreach to the LCA and rare inherited retinal disease community.

P.O. Box 705 | Ledyard, CT 06339



NON PROFIT ORG  
US POSTAGE PAID  
MYSTIC, CT  
PERMIT NO. 16