SOFIA SEES HOPE

Supporting the LCA and rare retinal disease community.
We see a cure for blindness.

SEEING HOPE | Newsletter

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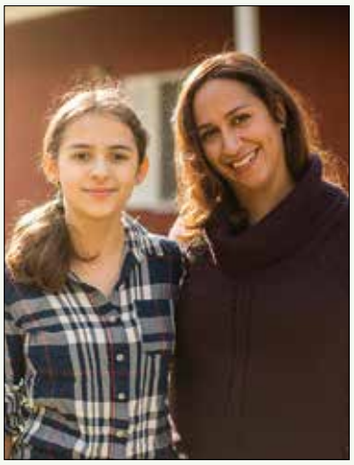
April 2019 | Issue 6

From the Founder

Sofia Sees Hope is a rare disease patient advocacy organization. We provide education and support for the Leber congenital amaurosis (LCA) patient community, and for other rare eye diseases. We raise funds for research, and to support free genetic testing and counseling. Since our founding in 2014, we have given $275,000 to research and more than $105,000 to provide families free access to genetic testing.



Sofia Priebe & Laura Manfre

We also put a lot of energy into finding and sharing patient stories. These stories allow our families to connect and learn from each other and hopefully feel less isolated. They give individuals a voice, and help increase general awareness of retinal disease, empowering our community and ultimately accelerating the path to treatment.

Usually you'll read the stories we share online or in our newsletter, but in March we did something a little different, and hosted an event we called A Rare Opportunity. We were so thrilled to welcome bestselling author Nicole Kear and America's Got Talent Golden Buzzer winner Christian Guardino to share their stories.

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Living with LCA: 'Know It's Not Something You're Going to Conquer in One Day'

Gunner Lincoln

By Rosanne Smyle

The best of both worlds—that's how Ashlyn Lincoln describes life with her two sons: 4-year-old Gunner, who was born without vision, and 7-year-old Ace, who is sighted.

"Both Ace and Gunner teach us many life lessons, regardless of who is sighted and who is blind," says their 29-year-old mom.

Living in eastern Iowa, Ashlyn and her husband, Axel, noticed problems with Gunner's eyes in August 2014, when he was about 2 weeks old: He stayed awake during the day and his eyes would not focus. The pediatrician examining their infant at 6 weeks wasn't concerned, but to put them at ease, he referred them to the first in what would become a series of specialists, leading to lots of tests on Gunner's eyes and on his parents' genetic backgrounds.

Gunner was diagnosed with Leber congenital amaurosis (LCA) at 4 months; at 6 months in February 2015, his parents learned he had LCA10, caused by a mutation in his CEP290 gene. Doctors determined Gunner came into this world with no usable vision cells and no light perception.

Last May, the family moved to a suburb of Nashville, seeking a stronger support system and better resources for Gunner, who is now thriving at the Tennessee School for the Blind.

Gunner loves prekindergarten, especially gym time and swimming lessons. He listens to try to understand musical instruments, and he loves making art and writing on the Brailler. "He's pretty independent," Ashlyn says.

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From the Founder

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At the age of 19, Nicole was diagnosed with retinitis pigmentosa, and told she would be blind in 10 years. She decides to make the most of the vision she has left. She joins circus school, tears through boyfriends, travels the world, and through it all, keeps her vision loss a secret. Her memoir, "Now I See You" is told with humor and irreverence.

Christian was diagnosed as a toddler with LCA2, a mutation in the RPE65 gene. Five years ago he participated in the clinical trial that led to the creation and approval of LUXTURNA™, the first gene therapy in the U.S. to treat genetic disease. He regained an enormous amount of vision. In 2017, he wowed the nation with his singing on America's Got Talent, was Howie Mandel's Golden Buzzer winner, and made it to the semi-finals.

Sharing stories like Nicole's and Christian's helps us make a difference. We know that losing your sight is challenging. We know that having a rare disease is hard. Despite this, from the moment we created Sofia Sees Hope, we have chosen the glass half full. We acknowledge challenges, and we always look forward. When we think of the people we have met, the science we have supported, the issues we have faced, and the stories we have shared—to us, it has all been a rare opportunity.

Thank you for supporting us in this journey. We couldn't do it without you.

Laura

In January 2014, Laura Manfre co-founded Sofia Sees Hope with her husband, Chuck Priebe, and their friend, Elisse Rosen. The nonprofit funds research to treat blindness caused by LCA and other rare IRDs and provides outreach, support, and advocacy for the patient community.

Upcoming Events

International Congress of Research on Rare and Orphan Diseases
Re(Act) Congress America
May 8–11 • Toronto, Canada
react-congress.org
The International Congress of Research on Rare and Orphan Diseases brings together research leaders and young scientists from a variety of breakthrough scientific fields to present cutting-edge research and exchange ideas. Patients and patient organizations committed to research also share their experiences and perspectives.

Global Genes RARE on the Road
Rare Disease Leadership Tour
May 4 • Birmingham, AL
May 18 • Denver, CO
July 13 • Sioux Falls, SD
globalgenes.org/event/rare-on-the-road
The EveryLife Foundation and Global Genes partner for RARE on the Road, a Rare Disease Leadership Tour, to bring education and insights to rare disease patients, caregivers, and other advocates.

Orphan Disease Center & Global Genes
RARE Drug Development Symposium
June 6–7 • Philadelphia, PA
• orphandiseasecenter.med.upenn.edu/events
• globalgenes.org/event/advocacy-symposium
The Orphan Disease Center at the University of Pennsylvania School of Medicine, in partnership with Global Genes, hosts the RARE Drug Development Symposium to connect, educate, and inspire rare advocates. The Symposium focuses on the drug development process and the role of rare disease advocates.

National Organization for Rare Disorders
2019 Living Rare, Living Stronger NORD Patient & Family Forum
June 21–23 • Houston, TX
rarediseases.org/living-rare-forum
During this inaugural event, the rare community will come together with physicians, medical students, and allied health professionals for a program of learning, sharing, and connection in a fun atmosphere of support and understanding. The gathering also features the Rare Impact Awards.

Foundation Fighting Blindness VisionWalk
Nationwide / ongoing
www.fightblindness.org/site/SPageNavigator/VisionWalk_Homepage
VisionWalk has raised more than $51 million to fund sight-saving research since its 2006 inception. Join the tens of thousands of people who have taken important steps toward a cure by taking part in a 5K VisionWalk. Find a walk near you and register today.

Do you have an event you want to share? Let us know! Email Rosanne@sofiaseeshope.org with the information and a link.

Living with LCA: Take Baby Steps

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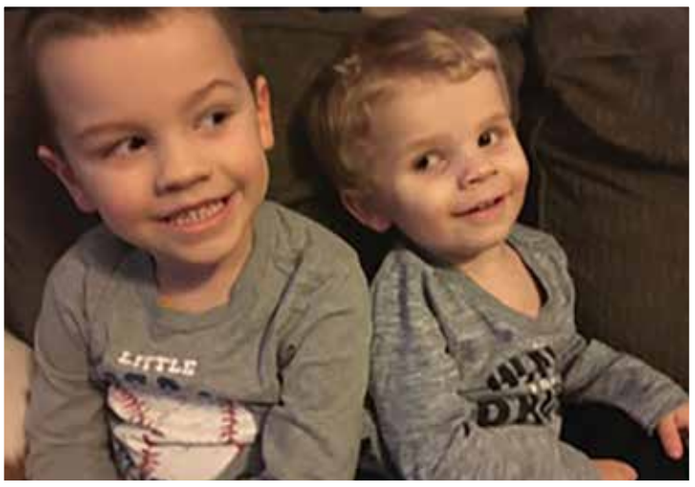


Ace and Gunner Lincoln

Ace and Gunner have a typical sibling relationship, blaming each other, kicking each other, playing in their own world, she says. "Ace pretty much treats him like he is sighted. He adjusts (when he remembers Gunner cannot see) and goes right back to thinking it. It's the best of both worlds.

"Their positive outlook and attitudes on life really help us be better ourselves. I just feel so lucky to be able to always have different outlooks on everything that others may not realize," she says.

Their daily life is like other families—the boys get up and get ready for school, eat breakfast, brush their teeth. After school they have snack, play, watch cartoons, do homework.

"This might be the only thing 'different,' " Ashlyn notes "Ace's homework is reading, so Gunner will bring his Braille books to the couch and 'read' them, too. Gunner's reading is feeling the Braille but making up his own stories as he goes, which are usually pretty creative and cute."

Ashlyn's best-of-both-worlds' concept might as well extend to the best of three worlds, given the influence, support, and love she and her family have received from Axel's "battle buddies" from his time in the Marines.

Axel served four years' active duty (followed by four years' inactive duty), being deployed twice to Afghanistan and once to Haiti on a relief mission.

He came home from Afghanistan suffering from severe injuries when his right shoulder took the brunt of his Humvee's impact after it ran over an IED, an improvised explosive device.

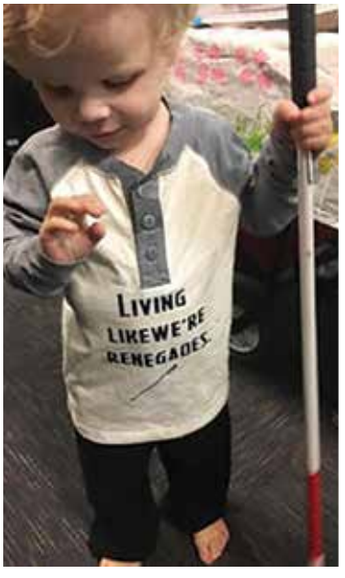
Axel and Ashlyn married as soon as they could after his return home. He has a 90 percent disability rating for the injury and for Post-Traumatic Stress Disorder (PTSD). About two years ago, Alex's service dog, Tucker, became part of the family, helping with his PTSD and bringing comfort when his anxiety is high. Tucker's also great with the boys and may someday be joined by a service dog for Gunner.

Dealing with Gunner's diagnosis and coping with new realities was difficult. Here is the message Ashlyn would like to extend to other mothers: "I was overwhelmed, and I was tired. I want to acknowledge that I know you're overwhelmed and it's OK.

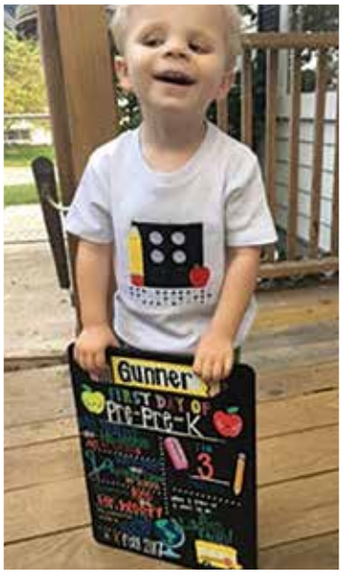
"Take baby steps, know your local resources, and know that it's not just something you're going to conquer in one day."

For support and to learn more about the LCA community, Ashlyn and a friend she met through an LCA Facebook group, traveled to Connecticut last fall to attend Sofia Sees Hope's LCA Family Conference, where she found a sense of community.

"It was fantastic," she says. "There are families in other states and I'm not alone and here we are together. It's just a moment where you can find comfort and know you're not alone."



Gunner with his cane



Gunner loves pre-kindergarten

Ashlyn says she does have a personal goal—one that she's fulfilled right here.

"It's hard to admit when your child is born and not perfect, and your husband has PTSD," she says. "I hope that just by telling my story about LCA and veterans, that other families also can not feel so alone."

ProQR's Clinical Trial for CEP290 Treatment: 'Great News for People with LCA10'

By Ben Shaberman
Senior Director of Communications
Foundation Fighting Blindness

ProQR, a biotech company in the Netherlands, reported vision improvements for patients in a Phase 1/2 clinical trial for QR-110, a therapy for people with Leber congenital amaurosis caused by the p.Cys998X mutation in the CEP290 gene (LCA10). The company said 60 percent of subjects in the Phase 1/2 trial demonstrated improvements in visual acuity and their ability to navigate a mobility course. The treatment was also safe.

Because of the encouraging interim results, ProQR has concluded the 10-participant Phase 1/2 trial and is preparing to move the treatment, now called sepofarsen, into a Phase 2/3 clinical trial in the first half of 2019.

Known as ILLUMINATE, the Phase 2/3 trial will be a randomized, double-masked, sham-controlled trial, initially enrolling 30 adults and children assigned equally to three parallel arms (two active dose levels and a sham control arm) with 10 participants in each arm. (In a sham control, the doctor goes through the motions without actually performing the treatment.)

The primary efficacy endpoint for the Phase 2/3 trial will be change in visual acuity from baseline in the treated arm compared to sham-treated control arm at the 12-month time point. Sham-treated participants may be offered cross-over to active treatment after 12 months, and all participants will continue to receive treatment for a total of 24 months after which they may be offered participation in an open label extension trial. (An open-label trial is a type of clinical trial in which both the researchers and participants know which treatment is being administered.) Treatment of the second eye will be adaptively incorporated into the trial.

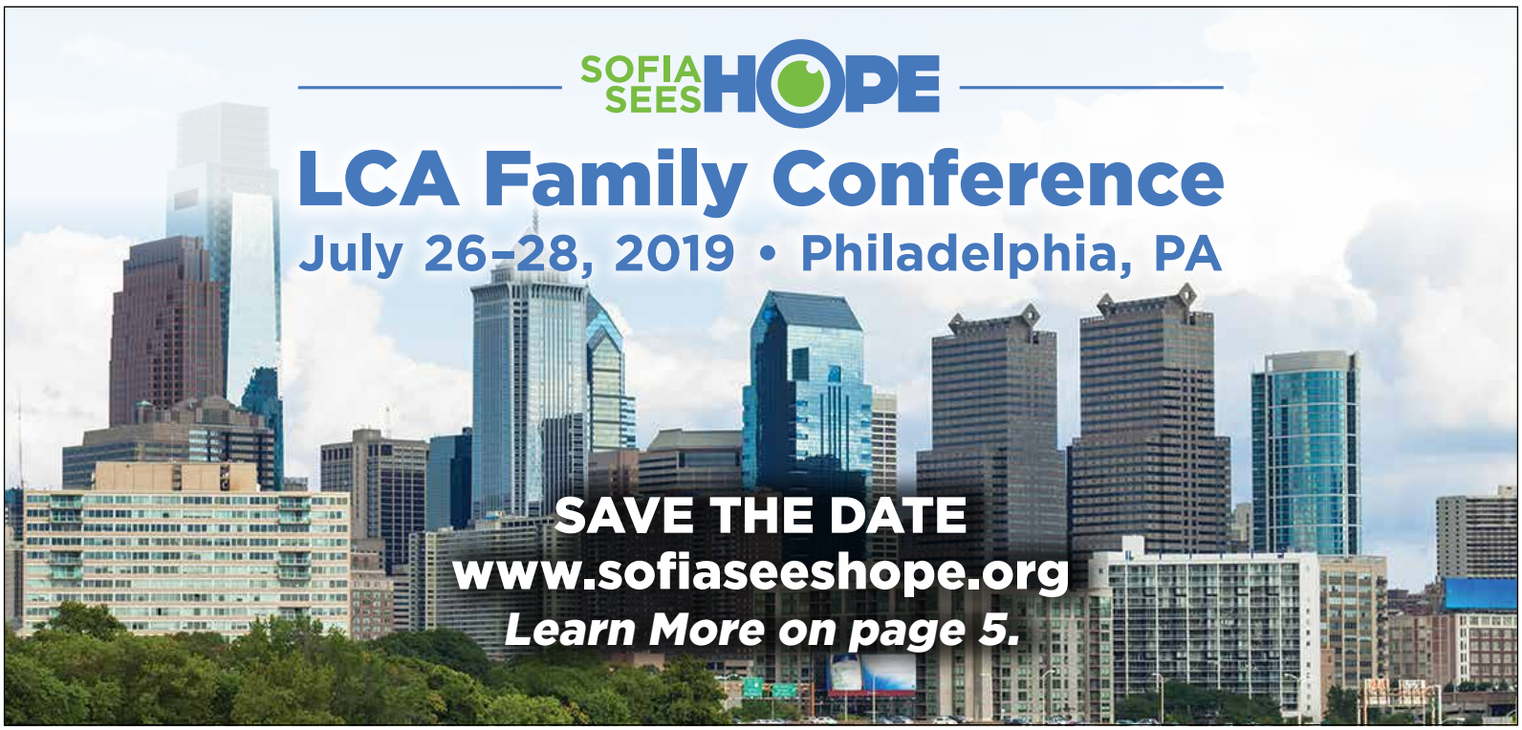
"The results from ProQR's Phase 1/2 trial are great news for people with LCA10," says Ben Yerxa, Ph.D., chief executive officer at Foundation Fighting Blindness. "Furthermore, the safety and vision improvements observed in the study provide strong evidence that antisense oligonucleotides, ProQR's treatment approach, are an effective way to save and restore the vision of people with retinal diseases.

"ProQR's results for QR-110 are groundbreaking, because it is the first time this treatment approach has improved vision in humans with retinal disease," he said.

ProQR's LCA10 therapy, antisense oligonucleotide (AON), works like "genetic tape" to repair the mutation. Unlike gene replacement therapies in which copies of whole genes are delivered to replace defective copies, AONs correct the mutation in the patient's messenger RNA, which conveys genetic information for protein production. AONs can be advantageous when large retinal-disease genes—such as CEP290 or USH2A—exceed the capacity of viral gene-replacement delivery systems.

The company plans to report top line results for the Phase 2/3 study around year-end 2020.

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



Navigating Adulthood by Jack McCormick

The Technology Is Out There To Help You Move Through Life

TRANSPORTATION

If you have a visual impairment and own an iPhone and haven't purchased **BlindSquare** yet, stop reading and buy it now. I promise that it will be the best $35 you ever spend. BlindSquare is a navigation app that announces your address, street intersections, and points of interest as you walk. It also has the ability to track locations, which is a feature I use every time I go somewhere new.

If you live in a community with public transit, I also recommend an app like **Moovit**. I use Moovit to get turn-by-turn directions to new bus stops, and there is also a feature where the app will notify you when it is time to get off the bus. This is especially useful when the bus is too loud to hear the overhead announcements.

PICTURE IDENTIFICATION

Depending on your level of vision loss, there are many apps that can help you read paper documents and identify objects. If you still have a fair bit of usable vision, there are many magnification apps from which to choose. I recommend downloading a few and picking what works best.

Seeing AI Free App

If you are like me and have very little usable vision left, download **Seeing AI**, a free app from Microsoft. If you want to see it, this app will help. You can identify colors, scan product barcodes, read print on paper documents, and get detailed descriptions of pictures from your camera or social media.

COOKING

I enjoy cooking but it doesn't come without its challenges. One of the biggest challenges I had when I started cooking was telling if meat was cooked through. That was until I got a **talking meat thermometer** from the Braille Superstore. Now my steak is always a perfect medium rare!

Jack McCormick graduated in 2018 from Canada's Wilfrid Laurier University in Waterloo, Ontario. Jack was diagnosed in high school with LCA2. He is a Sofia Sees Hope ambassador, helping people living with LCA and IRDs. You can read his blog at ***jackdamccormick.wordpress.com***

Get Set to Connect

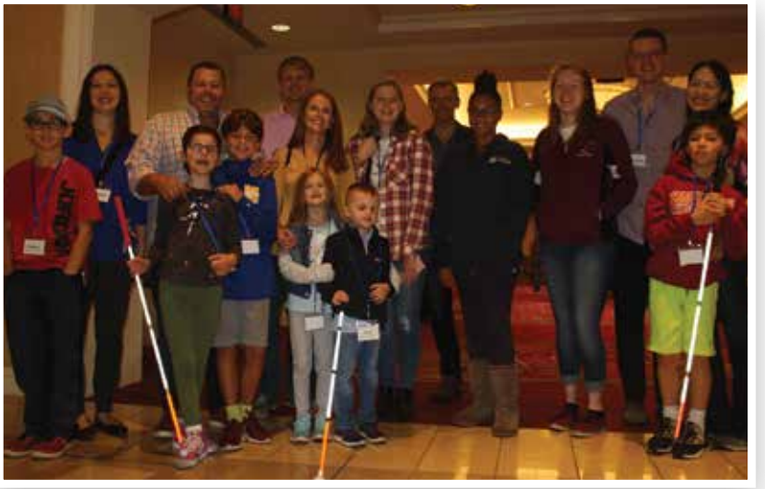
Sofia Sees Hope's second LCA Family Conference will be July 26–28 in beautiful downtown Philadelphia, at the Warwick Rittenhouse Hotel.

"The goal of this conference is to get people excited about advances in research, deepen your understanding of the roles various organizations play in developing treatments, and provide insight into how an active patient community can support and accelerate treatment," said Annette Tonti, executive director of Sofia Sees Hope.

At the conference, we will be covering the latest updates in Leber congenital amaurosis (LCA) and other inherited retinal disease (IRD) research. There will also be panels on the role of patients, the importance of the patient voice, and advocacy.

Most importantly, the conference will enable conversations for families and caregivers, explore issues associated with living with IRDs from childhood to adulthood, and give patients and families a place to connect, to learn, and to create lasting relationships with others.

The conference will include leaders from industry, research organizations, and government, as well as families and patients affected by IRDs. The full schedule and list of speakers can be found at: www.sofiasees.org/event/lca-family-conference.



In 2018, Sofia Sees Hope hosted its first LCA Family Conference in Groton, CT. About 60 people attended from around the country and Mexico.

"I attended the (2018) conference, and thanks to them I was able to meet a girl with the same genetic mutation that I have (the first I know around the world)," said Angélica Bretón Morán, who traveled to the conference from her home in Mexico. "And I was able to chat in person with the doctor who almost three years ago gave us my genetic result by phone. It was a huge blessing for me and for my family, it means a lot, more than I can explain."

CT Rare Disease Day 2019: Sofia Sees Hope Promotes Accessibility to Treatment

By Elissa Bass

Participants in Connecticut's Rare Disease Day event

Sofia Sees Hope celebrated Rare Disease Day 2019 with Connecticut lawmakers, urging them to preserve patient access to approved treatments.

Rosanne Smyle, a staff member of Sofia Sees Hope, joined more than two dozen speakers on February 28 for an informational session that included legislators, patients, doctors, researchers, nurses, caregivers, advocates, advocacy organizations, and business leaders.

"Our state legislature needs to know that we fully support the principle that all FDA-approved treatments should be made available to all those who will benefit from such treatment, and to reject any proposed requirements restricting access to medications," Smyle said.

In her introduction, Smyle told the gathering of about 130 people at Hartford's Legislative Office Building 2nd Floor Atrium that Sofia Sees Hope advocates for patient access to resources and treatments to help transform the lives of those affected by blindness caused by rare inherited retinal disease. She told them Leber congenital amaurosis, known as LCA, is one of the more than 7,000 rare diseases affecting 300,000 people in Connecticut and 30 million nationwide.

Along with Connecticut's celebration, more than 30 advocacy events took place at state Capitols and other locations throughout the United States. Globally, more than 90 countries celebrated Rare Disease Day, held annually the last day of February. Rose Avellino, Grassroots Advocacy Manager for the National Organization for Rare Disorders (NORD), and Lesley Bennett, Connecticut's Volunteer State Ambassador for NORD's Rare Action Network, hosted and organized Hartford's event.

In honor of the day, Connecticut Gov. Ned Lamont signed an official proclamation that said, in part: "Whereas, individuals and families affected by rare diseases often experience problems such as diagnosis delay, difficulty in finding a medical expert, and lack of access to treatments and services," and "Whereas, thousands of patients and caregivers, medical professionals, researchers, medical companies, nonprofit organizations, and others in the state of Connecticut will participate in that observance, I, Ned Lamont, Governor of the State of Connecticut, do hereby proclaim February 28, 2019, as Rare Disease Day in the State of Connecticut."

The statehouse group learned from Sofia Sees Hope that LCA is characterized by severe vision loss at birth.

"While some children are born with little or no vision, others may have significant vision loss in the first few years of life, stable vision for a period of time, and then eventually complete vision loss as the retina deteriorates into total blindness," Smyle said. "More than 25 genes are associated with LCA and a mutation in just one of these can result in blindness."

A great source of hope for improving vision emerged in the LCA and Inherited Rare Disease (IRD) community with the development of LUXTURNA™ by Spark Therapeutics and the medication's 2017 Food and Drug Administration approval. After decades of research and dedicated

investment into studies, Smyle said, scientists created this ground-breaking genetic therapy that helps restore vision in patients with LCA2 or RPE65, one of the genetic mutations causing LCA. LUXTURNA™ also is the first genetic therapy ever in the United States to treat any inherited rare disease.

"LCA patients treated with LUXTURNA experienced dramatic changes in their lives with greatly improved or restored vision. Five, 6, 7-year-old children treated with LUXTURNA view life in a new light in big and little ways," Smyle said. "They now can see rainbows arcing in the sky and stars shining at night."

She told the group that the optimal window for reversing vision loss is during the early phase of the disease.

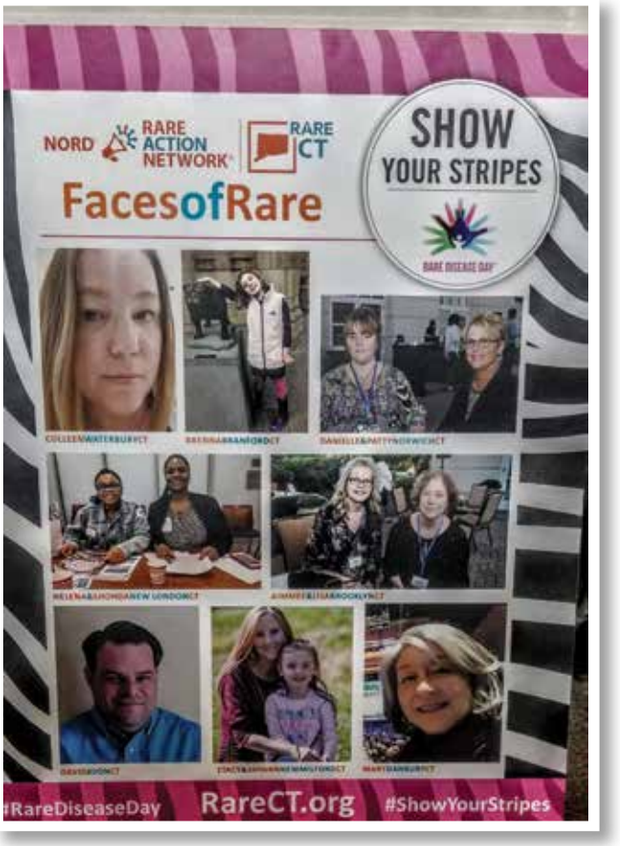
"Unfortunately," she said, "several states considered laws requiring a certain degree of blindness before treatment—requirements that go against federal health recommendations and fly in the face of basic humanity."

Rosanne Smyle speaking at RDD event

Connecticut has not yet had to deal with this kind of legislation, but Sofia Sees Hope wanted lawmakers to know that requirements limiting access or withholding treatment increase the potential for failure and for the likelihood of the disease recurring.

Smyle also told the group that more money is needed for treatment research and that since its founding five years ago, Sofia Sees Hope raised and donated $275,000 for genetic retinal research, genetic testing and counseling that is vital to move treatment forward. There are 23 gene-based clinical trials targeting 13 genes, and more than 20 retinal cell therapy trials in progress. Another 100 genes are under investigation in the preclinical pipeline.

Sofia Sees Hope also encouraged the Connecticut General Assembly to establish a Rare Disease Advisory Council comprised of legislators, patients, patient advocates, doctors, researchers, business leaders, and community members to address the emerging public health priority of rare diseases, including LCA and other IRDs.



Want to Get Involved with Rare Disease Activities Where You Live?

There are many organizations acting locally and globally, here are a few to start with!

In the US:
National Organization of Rare Disorders
www.rarediseases.org

In Canada:
CORD
www.raredisorders.ca

Internationally:
Rare Diseases International
www.rarediseasesinternational.org

SAVE THE DATES

Do you have an event you want to share? Let us know! Email Rosanne@sofiaseeshope.org with the information and a link.

Global Genes 2019 RARE Patient Advocacy Summit

September 18–20 • San Diego, CA • globalgenes.org/2019summit

The RARE Patient Advocacy summit, a can't-miss event of the year for rare disease stakeholders, is the largest gathering of rare disease patients, advocates, and thought leaders worldwide. Join Global Genes, partners, and rare disease community members to become equipped with the tools needed to persevere and thrive.

Sofia Sees Hope Dinner in the Dark

October 19 • Groton, CT • sofiasees.org/get-involved/dinner-in-the-dark

Save the date for the Sofia Sees Hope 6th Annual Dinner in the Dark. Our primary fundraiser, this gala event at the Mystic Marriott is not to be missed. Diners enjoy a multi-course gourmet dinner paired with fine wines, followed by live music and dancing.

National Organization for Rare Disorders (NORD)

Rare Diseases and Orphan Products Breakthrough Summit

October 21–22 • Washington, D.C. • rarediseases.org/summit-overview

National Organization for Rare Disorders' annual Breakthrough Summit brings together more than 800 leaders from the Food and Drug Administration, National Institutes of Health, industry groups, payers, and research institutions. The summit features networking with today's rare disease innovators, cutting-edge keynote speakers, and six breakout sessions.

THIS NEWSLETTER IS MADE POSSIBLE BY THE GENEROSITY OF:

- Spark Therapeutics
- Sanofi Genzyme
- MeiraGTx

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

To learn more about Sofia Sees Hope visit our website at 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.

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