

SEEING HOPE | Newsletter

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September 2020 | Issue 9

From the Founder Laura Manfre

2020: Making the Best of It



*Laura Manfre, Orly Shamir & Sofia Priebe
at Dinner in the Dark 2019*

If you're a believer in creating a vision or mood board to set the tone for your upcoming year, I'm guessing you've long ago thrown out the one you made in January 2020.

Here we are, three-quarters of the way through a year that no one could have predicted. Back in December, we were busy tabulating our 2019 revenue and making our year-end donations to research projects and to ensure families could access free genetic testing. We gave \$85,000 to four research projects, and more than \$20,000 to provide free genetic testing through the My Retina Tracker® program, an open-access, no-cost genetic testing program for individuals living in the U.S.

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The ABCs and 123s of Jordynn

By Rosanne Smyle

Blink and you just might miss Jordynn rocketing past you.

A force to be reckoned with when it comes to music and movement, Jordynn is 4 years old and lives with Leber congenital amaurosis (LCA) caused by a mutation in her RPE65 gene.

"Anything that moves, Jordynn is willing to try. The swings," said her mother, Joy. "We have a hard time getting her off the swings. At school, they use it as a reward for what she accomplished."

Jordynn and her mother, who live in upstate New York near Rochester, received their genetic diagnoses in 2017, just months before the U.S. Food and Drug Administration approved LUXTURNA®, a genetic therapy for patients with the RPE65 gene mutation, known as LCA2.

Jordynn was on a waiting list to take part in an RPE65 gene therapy study by Dr. Mina Chung of the University of Rochester Medical Center (URMC) Strong Flaum Eye Institute when Dr. Chung died in February after a fall while skiing in Italy.

Joy is waiting to hear when Jordynn can join the study and was told by Dr. Benjamin Hammond, a colleague of Dr. Chung's and an ophthalmologist working with Jordynn, that everything is on hold until another surgeon comes on board.

Jordynn celebrated her 4th birthday May 6. With her prekindergarten class closed because of coronavirus, Jordynn had her schoolwork sent home and through a YouTube

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*Seated: Jordynn, her mom Joy, grandma
Gwen. Standing, aunt Jackie at the
2019 LCA Family Conference.*

The ABCs and 123s of Jordynn

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channel. She had music therapy incorporated with orientation and mobility training because she is so drawn to playing the piano and the drums and loves to follow the beat and sing.

She also is learning braille on a Braille, and everything throughout her mom's apartment and her grandmother's nearby home is labeled in braille for her to identify.

"Before it was all about Sesame Street," Joy said of her daughter's learning. "Right now, it's ABCs and 123s and colors and all the things on YouTube that she can dance to and learn her numbers while she's singing."

PRAISE FOR NEW YORK'S RESOURCES

Jordynn's vision allows her to follow light and see three-dimensional shapes, but she cannot see them on paper.

Her mother first noticed something might be awry with her vision when she wouldn't pay attention to people looking at her and smiling. Doctors diagnosed Jordynn with nystagmus, a vision condition wherein the eyes make repetitive, uncontrolled movements.

With a later diagnosis of LCA, Joy was stunned. "I just sat there, and then I said, is this my fault? Is there something I could have done when I carried her? OK, it's something genetic."

"I cried," she said. "The whole family cried."

Already working with URMIC Pediatrician Dr. Mary Porter for early-mobility intervention, Jordynn's family found more resources through Dr. Chung and Dr. Hammond, and through upstate New York's Association for the Blind & Visually Impaired.

Through it all, though, Joy is most grateful for an incredibly supportive family. Jordynn's family support extends to her dad in North Carolina, aunts, uncles and four grandparents, including Grandma Gwen, Joy's mom.

"It's just family support; that's how we get through this," Joy said. "Family support and lots of toys."

Jordynn is among Gwen Goodwine's 13 grandchildren and gets to see her grandma almost daily because



Gwen Goodwine

they live near each other and because Gwen takes care of her while her mom, Joy, cares for the elderly and those with dementia.

There is no disguising the abundant love and hope that Gwen exudes for this grandchild. For Jordynn, Gwen set up rugs like oversized dominoes throughout her house, blue and white rugs, from the family room, to the living room, to the kitchen, to the bedroom.

"That's how she learned how to navigate," Gwen said. "She runs through here like she's got 20/20 vision."

"She'd jump off this house if she could. I bought her a rocking horse. She'll get on that horse and say, 'I'm going to Tennessee, I'm going to Georgia, I'm going to California.' She tore it up. Got another one. How she used it! They sent us another one free."

Gwen, now 80 with still a lot of energy, wants more than anything to see Jordynn see.

"I say, Lord, please. I don't want to leave this earth 'til she look at me and see my white hair, take both of her hands on my jaws and kiss me."

MAKING IMPORTANT CONNECTIONS

After Jordynn's genetic diagnosis, Joy and Gwen found out about Spark Therapeutics, the research company that developed LUXTURN[®], the first and only approved gene therapy for inherited disease in the United States and Europe. The drug is an engineered virus that delivers the human RPE65 gene by subretinal injections.

They spoke with Spark's then-Head of Patient Advocacy, Jamie Ring, who told them about Sofia Sees Hope. Joy, her sister Jackie, her mom, and Jordynn attended Sofia Sees Hope's LCA Conference in July 2019.

The many people they met included Dr. Jean Bennett, who with her colleagues at Children's Hospital of Philadelphia and Spark Therapeutics, developed LUXTURNA®. They also spoke extensively to Ben Shaberman, Senior Director of Scientific Outreach and Community Engagement for the Foundation Fighting Blindness.

Joy said it was great to meet families with kids who have gone

through the same experiences. "Learning about the treatment and getting the education about all of it really gave me something to think about as my daughter's journey continues as she lives with this visual impairment," she said. "Knowing that my daughter can thrive and live a happy life with some occasional bumps in the road was a wonderful feeling."

She's learned the importance of perseverance and patience,

offering this advice: "Don't get discouraged. Take your time. Learn the process. Get to know your options. Find the resources available in your state. It's a process but it takes time and sometimes it can be frustrating."

Joy's other message? "Please treat Jordynn like a normal toddler, because this is her normal and she is just like any other toddler."

2020: Making the Best of It

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We were planning lots of programming and events for the coming year, including of course our signature Dinner in the Dark, a Rare Opportunity with best-selling young adult author Kody Keplinger, who lives with LCA, and other local fundraisers.

And then, coronavirus arrived. In addition to the millions of people infected, the hundreds of thousands who have died, and the countless lives turned inside out by the ripple effects of the virus across the globe, this pandemic has had a chilling effect on research around anything except a cure for COVID-19.

It's heartbreaking for those in the IRD community to know that incredible work—either in clinical trials, or on the cusp—was put on hold while research turned to a coronavirus vaccine and/or treatments. It's understandable, but it's still tough. The good news is, that is turning around and research, including clinical trials, is picking back up.

While it's hard to believe we will get to the other side of this crisis any time soon, we will get there, and so we have been working to make sure that Sofia Sees Hope remains viable, vibrant, engaged, and active. We have created virtual events designed to engage our audience and fundraise simultaneously, including a fun Socially Distanced Scavenger Hunt we did online in April that attracted 32 teams from around the globe to complete a series of tasks. We raised more than \$5,000 and made some really good new friends. We also Zoomed with master mixologist Dale DeGroff about martinis, and sold autographed copies of his new book, raising more than \$10,000.

And instead of Dinner in the Dark, we are holding Bidding in the Dark, coinciding with World Sight Day on Oct. 8. This will be both mysterious and fun! Stay tuned to our website and social media for more information.

In some ways, learning to live during this pandemic is like those days right after you get that diagnosis of LCA, either for yourself or your child. You're stunned. Speechless. Maybe knocked down. But you come to grips with your current situation, and you pivot. You adapt and you move forward. You continue to live your life and look to the future. That's what we're doing, and we hope you are too.



Dinner in the Dark 2019 Event Committee

VISIONS 2020 Conference

Highlights Emerging LCA Therapies



By Ben Shaberman
Senior Director, Scientific Outreach & Community Engagement

FOUNDATION FIGHTING BLINDNESS

Like so many events this year, the international conference of the Foundation Fighting Blindness, VISIONS 2020, was held virtually to help contain the spread of coronavirus. But there was nothing “virtual” about the promising news on emerging therapies for LCA reported during the three-day, online event, which had more than 1,600 registrants.

For LCA families, the LCA Science and Research session was a highlight of the conference. Hosted by Eric Pierce, MD, PhD, director of the Berman-Gund Lab for the Study of Retinal Degenerations at Mass Eye and Ear (MEE), and Christine Kay, MD, a clinical researcher at Vitreo Retinal Associates in Gainesville, the 90-minute session provided a plethora of updates on the LCA research landscape.

“LCA is the most exciting field for me to be in because we’re finally able to bring basic science advances into the clinic,” said Dr. Kay during the session. “And one of them (LUXTRNA®) is FDA approved and commercially available.”

While Drs. Pierce and Kay discussed and referenced LUXTRNA® and the clinical trial that made it possible, the doctors gave updates on a variety of emerging treatments that are in, or moving toward, human studies.

RPGRIP1 (LCA6)

Dr. Pierce reported that an RPGRIP1 gene therapy, which has been under development in his lab for many years, has shown “beautiful proof-of-concept” in animal models and has been licensed to PTC Therapeutics for clinical development. The therapy will hopefully be in a clinical trial in two to three years.

NMNAT1 (LCA9)

Also in development at MEE is a gene therapy for patients with NMNAT1 mutations. Ten years ago, the lab found that mutations in the gene cause LCA, and subsequently developed an animal model that is similar to the human condition. After several years of research, the lab developed an NMNAT1 gene therapy that works in mice, and a paper on its success is forthcoming. They are now advancing the treatment toward a clinical trial.

CEP290 (LCA10)

Two projects underway for CEP290 were discussed during the LCA session, both of which target the p.Cys998X mutation.

ProQR’s RNA therapy—an antisense oligonucleotide (AON)—works like genetic tape to mask the mutation and is in a Phase 2/3 clinical trial. Known as sepfarsen, the emerging treatment improved vision in 60 percent of trial participants in the Phase 1/2 trial. Daniel de Boer, chief executive officer at ProQR, also presented information on sepfarsen during the session entitled “The Promise of Genetic Therapies.”

Editas, in collaboration with Allergan, is conducting a Phase 1/2 clinical trial for a CRISPR/Cas9 treatment (targeting p.Cys998X). CRISPR/Cas9 works like a pair of molecular scissors to cut out the mutation. No reports have come from the trial thus far. Dr. Pierce noted that this study is the first for evaluating CRISPR/Cas9 in the human body.

Dr. Pierce also commented that these techniques, if they work, could be applied to other common CEP290 mutations.

In the session “Emerging Technologies,” Hemant Khanna, PhD, at UMass Medical School, presented his work on a “mini-gene” CEP290 gene therapy, which is designed to address all mutations in CEP290. Iveric bio is a partner in his project. Mini-genes are an approach for CEP290 gene delivery, because the gene is otherwise too big to fit in the viral containers used for gene delivery.

CRB1 (LCA8)

Horama, a gene therapy development company in France, is licensing an emerging gene therapy for CRB1 mutations from the Leiden University Medical Center in the Netherlands. The goal is to launch a clinical trial of the treatment in 2023. Dr. Pierce noted that other groups in the United States are also working on CRB1 gene therapies.

GUCY2D (LCA1)

Shannon Boye, PhD, at the University of Florida, was the featured presenter in the Opening Session of VISIONS 2020. She covered a lot of ground in gene therapy during her talk, including her work in dual vectors for delivering large genes. Her lab’s efforts have also led to a Phase 1/2 GUCY2D gene therapy clinical trial at the University of Pennsylvania. The global pharmaceutical company Sanofi is sponsoring the study.

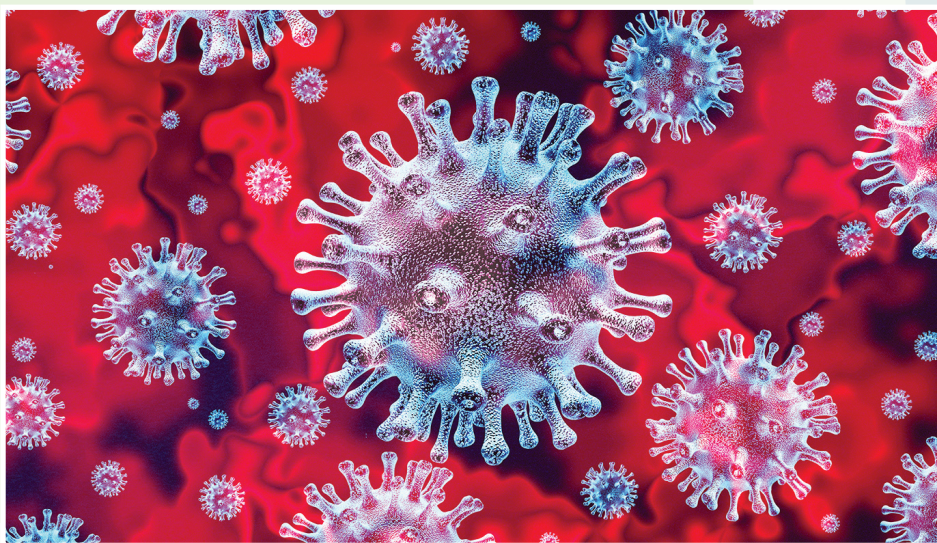
Recordings for all VISIONS 2020 sessions, including the LCA Science and Research Session, are available at: fightingblindness.org/recorded-sessions

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

Life in the Time of COVID

I first heard about coronavirus on my way back from a weekend of skiing in mid-January. It seemed to be the only thing the radio station we were listening to was talking about. At the time, COVID-19 was only reported in China and I didn't think it would ever impact my life. I've never been so wrong.

On March 13 I was scheduled to fly to Australia for a dream vacation with my childhood friend. I was subscribed to travel advisories issued by both the Canadian and Australian governments. I woke to an email from the Australian government advising against all travel.



I phoned my friend. We needed to cancel the trip. I don't think he appreciated the 5 a.m. call. We made the right decision. By noon Canada had issued its own travel advisory. I have never spent so much time on hold and hope to never again. Eventually I received refunds for the flight and hotels. This was only the beginning.

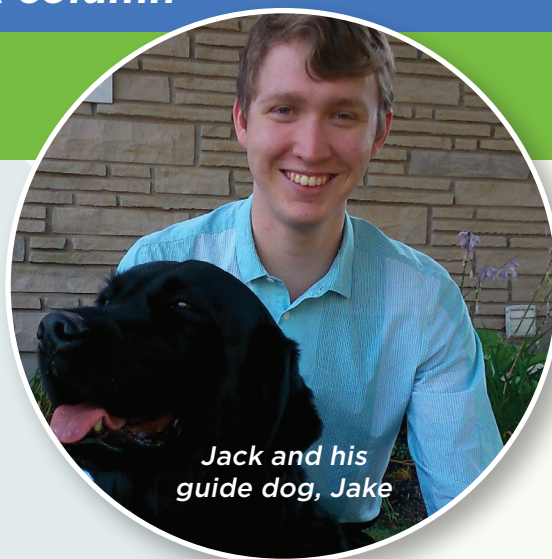
I work in the human resources department of a hospital. I spent the time I had planned to be on vacation working non-stop. We had to keep our patients

and staff safe. Government guidelines seemed to change every day. We didn't have enough staff. Everyone in our department was working three jobs.

I had no food. I had eaten everything—not wanting to return from Australia to rotten food. As someone with a visual impairment I get assistance from a store employee when I do my groceries. Getting this assistance meant being physically close to someone who wasn't part of my regular interactions—something I didn't feel comfortable doing, given the pandemic.

The alternative was delivery and it was near impossible to book a time. When I finally did, the shopper couldn't find most of the things I had requested because people were buying up everything. I soon ran out of food again. Someone from work offered to help me do my groceries. The shelves were empty. It felt like something out of a movie.

We fell into a new rhythm at work. I lost count of the number of people I hired. To limit the number of people in the



Jack and his guide dog, Jake

hospital, those who could work from home did. This included me most days. I hardly left my apartment.

Even now, there isn't much to do outside work. We are all struggling. We don't know when it will be over.

Throughout this experience, my vision loss has created a host of challenges. It has also helped me live in the pandemic. With decreasing vision, I have learned to adapt to change—something we have all had to do in 2020. My vision loss has made me stronger. Living through the pandemic will do the same for you. For me, I've learned more about pain, struggles and the power of connection. I've learned to acknowledge the challenges I experience and connect with the people who are important to me (even if it is virtually) to support each other because we are better together, even when we are far away.

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

EVENT CALENDAR - FALL 2020

Global Genes

LIVE! A RARE Patient Advocacy (un)Summit

September 14-25, 2020

Virtual Event

globalgenes.org/event/live

This two-week event provides members of the rare disease community, stakeholders, and allies the opportunity to connect and engage through interactive activities paired with educational programming. Participants will gain insight into the latest in rare disease innovations, best practices for advocating on individual and organizational levels, and strategic ways to accelerate change.

Sofia Sees Hope

Bidding in the Dark

October 8-10, 2020

Virtual Event

sofiasees.org/event/bidding-in-the-dark

We couldn't gather this fall to don our blindfolds, taste wine and indulge in a gourmet menu at Dinner in the Dark, our flagship event originally scheduled for October 10. So, our Event Committee brainstormed (over Zoom of course!) and came up with what we hope will be the next best thing. This event includes bidding on items to support Sofia Sees Hope, but we will not provide any visuals, and descriptions will be limited. Similar to Two Blind Brothers' "Shop Blind" campaign, we are asking our donors to trust us, and well, do more than just imagine buying a work of art that you cannot see. We will provide clues to the items, and we will also provide hints that participants can unlock by watching or listening to short videos and messages and answering a question about the content. The event opens October 8, World Sight Day. Stay tuned for more details!

National Organization for Rare Disorders

Diseases and Orphan Products Breakthrough Summit

October 8-9, 2020

Virtual Event

rarediseases.org/summit-overview

As technology in rare disease accelerates, and with topics like drug pricing being a top priority in government, we must act now to drive innovation and collaboration to develop stronger resources, better outcomes, and find cures for the community. Given the impact of the COVID-19 pandemic, connecting stakeholders and advancing meaningful dialogue is now more critical than ever to improving the lives of more than 25 million Americans living with rare diseases. Please join us for networking with rare disease innovators and for learning about building a stronger healthcare system from our blue-ribbon panel of public health experts, patient advocates, and industry leaders.

Foundation Fighting Blindness

Fall National Virtual VisionWalk

October 24, 2020

Virtual Event

fightingblindness.org/visionwalk

Join us for our Fall National Virtual VisionWalk to celebrate our Fighting Blindness community. While we may not be in the same places physically, we are joining across the nation to show the world what it means to fight blindness and be #VisionWalkStrong. We encourage you to take steps to support the Virtual VisionWalk by walking on a treadmill, taking a socially distanced walk outdoors, or even hosting a virtual meeting with your team to connect. VisionWalk, the Foundation's signature event, has raised more than \$55 million to fund sight-saving research since 2006. Together, we step closer to fighting blindness!

LCA Families 'Mixed & Mingled' at the Virtual VISIONS Conference

VIRTUAL VISIONS 2020

By Rosanne Smyle

Families living with Leber congenital amaurosis came together virtually, sharing their hopes and triumphs, their challenges and frustrations as part of the three-day Virtual VISIONS 2020 conference in late June, the annual flagship event sponsored by the Foundation Fighting Blindness.

Beth Borysewicz, Vice Chair of the Board of Sofia Sees Hope and a teacher of students with visual impairment, moderated the LCA Mix & Mingle forum that gave participants new ideas, new contacts and new knowledge about ongoing clinical trials and an approved LCA treatment.

A mother in Canada described her frustrating search for the right doctor and appropriate resources to help her 15-month-old son, who has LCA1 caused by a mutation in his GUCY2D gene. With months passing and her son growing, she worried that delays could impede helping her child.

She received specific contact information for a Canadian retinal specialist from Ben Shaberman, the Foundation's Senior Director for Scientific Outreach & Community Engagement. He also reassured her that she has not missed out on a GUCY2D clinical trial because it is not ending; it is moving into another phase and she would need to reach out to researchers conducting the trial.

Amy Reif said her daughter did not take part in the trials for the now federally approved LUXTRNA® gene therapy, but she described the girl's visual improvement two years after she underwent treatment for LCA2 caused by a mutation in her RPE65 gene.

"I will say that LUXTRNA® is not a cure," she said. "It's a treatment and things have improved immensely. It's not perfect, but it has really improved just the way she is able to get around and her sensitivity to light is so much better."

Krista Giannak, diagnosed with LCA-CEP290, tried to get into a clinical trial.

"I wanted to go for it," she said. "I have a sense of adventure and I thought it might be interesting to learn what color looked like or what art would be like or even just to have more light perception potentially."

But during the clinical trial screening, Krista learned she had more light perception than she previously had thought, but not enough to go forward in that particular trial.

A father of a toddler living with LCA shared that only one copy of the mutated gene NMNAT1 had been found and asked how to go forward, knowing both copies of the mutated gene have to be found for a confirmed genetic diagnosis.

The father also asked about the chances of a second child also having LCA. Laura Manfre, Co-Founder and President of Sofia Sees Hope, shared that her family had a similar concern after her daughter, Sofia, was diagnosed with LCA and that she understood his apprehension. She and her husband did decide to have another child, who is 10 now and does not have visual impairment.

During the virtual session, participants also discussed exchanging contact information to talk together in the future.

"Making connections!" Borysewicz said. "That's what this part of the conference is all about."

You can view the recorded sessions from the conference at fightingblindness.org/recorded-sessions

SAVE THE DATE

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

SOFIA SEES HOPE WE SEE A CURE FOR BLINDNESS.

BIDDING IN THE DARK

A two-day virtual auction
to benefit Sofia Sees Hope

October 8-10, 2020

Visit www.sofiaseeshope.org
for more info
and sponsorships.

Sofia Sees Hope LCA Family Conference 2021

July 9-11, 2021 • Cleveland, OH
sofiasees.org/events/category/lca-family-conference

Join Sofia Sees Hope in Cleveland for its third LCA Family Conference providing information and networking for the Leber congenital amaurosis community! Lots more details to come.



To learn more about Sofia Sees Hope, visit our website at www.sofiaseeshope.org.

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