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

SEEING HOPE | Newsletter

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LCA Family Conference 2019: Coming Together in Philadelphia

By Rosanne Smyle

The news is out from our 2019 LCA Family Conference, and it's terrific!

Families living with Leber congenital amaurosis (LCA) and other rare inherited retinal diseases (IRDs) learned at Sofia Sees Hope's July 27th conference in Philadelphia that they are living in a time of the most dramatic growth ever in genetic research.

But—as with most good things—there is a caveat: Patience.

While researchers report a record number of genetic studies in various stages, they face long and arduous journeys in developing federally approved treatments.

In her keynote address, Dr. Katherine High, co-founder, president and chief scientific officer of Spark Therapeutics, said initiation of a clinical trial to licensing of a product easily could take seven to 10 years.

"My take-home message is patience is a requirement in drug development." High said.

More than 80 people—patients, family members, advocates, doctors, researchers, and biotech industry leaders—gathered at the conference from July 26–28 in Philadelphia.

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Dinner in the Dark 2019: Come Take Flight(s) With Us

By Elissa Bass

There's no laurel-resting for the committee behind Sofia Sees Hope's annual Dinner in the Dark—this year the event has added flights of beer or wine to the menu, as well as an incredible new Majestic Box fundraiser!

That's right, the 6th annual Dinner in the Dark promises to be bigger and better than ever. Mystic Marriott Hotel and Spa Executive Chef Mark Vecchitto has planned a sublime gourmet three-course menu to be sampled while blindfolded, followed by the ever-popular dessert buffet. The evening begins with a cocktail reception at 5:30 PM, dinner at 7:00 PM, followed by a live auction and dancing.

Proceeds go directly in support of our mission to fund research for rare genetic disease and support our outreach and education work for the patient community. This event is a true culinary adventure and has become a regional must-attend!

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Dinner in the Dark

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New this year: When you purchase your ticket, you can choose to enjoy a flight of wine, courtesy of Angelini Wine, or a flight of beer, courtesy of our newest sponsor, Outer Light Brewing Company! Beer flights are limited to 100, so don't wait if you prefer a cool brew.

Once again, our sponsor Simply Majestic has gone above and beyond to create a magical piece of jewelry for our Majestic Boxes. Inspired by the zebra, the international symbol of rare disease, this year's pendants are designed exclusively for Dinner in the Dark.

Why is the zebra the symbol for rare disease? Medical students are trained to look for the most common diagnosis when a patient presents. In the 1940s, Dr. Theodore Woodward, a professor at the University of Maryland School of Medicine, told his students, "When you hear hoof beats, think of horses, not zebras." Dinner in the Dark raises funds to treat rare inherited retinal disease, so we say, show off your stripes!

Thank you to our generous sponsors, without whom this event would not happen:

- ABC PhotoLab
- Andersen Oil Company
- Angelini Wine
- Design Intervention, LLC
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- Editas Medicine
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- Window of Giving

Visit sofiasees.org/get-involved/dinner-in-the-dark/ for more information and to purchase tickets, Majestic Boxes, or become a sponsor.

Upcoming Events

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 for rare disease stakeholders, is the largest gathering of rare disease patients, advocates, and thought-leaders worldwide.

Sofia Sees Hope 6th Annual Dinner in the Dark

October 19 • Groton, CT
sofiasees.org/get-involved/dinner-in-the-dark
This gala event, our primary fundraiser, helps fund research to cure blindness caused by LCA, provides support for genetic testing and drives awareness, education, and connections for LCA and IRD families. This is a lively, sensory adventure and a true culinary event offering a unique menu and wine and beer flights, followed by live music and dancing.

National Organization for Rare Disorders (NORD)

Rare Diseases & Orphan Products Breakthrough Summit
October 21–22 • Washington, D.C.
rarediseases.org/summit-overview
The summit brings together more than 800 leaders from the U.S. Food and Drug Administration, National Institutes of Health, industry groups, payers and research institutions. The summit features six breakout sessions and keynote speaker FDA Acting Commissioner Norman E. "Ned" Sharpless. Topics include drug pricing, advancements in technology, and more.

World Orphan Drug Congress Europe 2019

November 12–14 • Barcelona, Spain
terrapinn.com/conference/world-orphan-drug-congress/index.stm
World Orphan Drug Congress Europe presents Strategy, Advocacy, and Partnering for the Orphan Drug Industry. The gathering brings together industry pioneers to share their insights on their innovative work across the whole value chain. Featuring more than 200 speakers, the congress offers opportunities to network, discover, and initiate life-changing discussion. Topics include clinical development, advocacy, and global patient and market access.

LCA Family Conference 2019

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The conference was sponsored by MeiraGTx, Editas Medicine, Spark Therapeutics, Sanofi Genzyme, Foundation Fighting Blindness, Allergan, ProQR, Two Blind Brothers, Applied Genetic Technology Corp. (AGTC) and Lions Clubs International.

Laura Manfre, the co-founder of Sofia Sees Hope and president of its Board of Directors, said the nonprofit's second family conference was designed to bring information to patients and families, as well as connect those patients to researchers and industry.

"A huge part of our mission is to make sure that members of the rare disease community do not feel alone," Manfre said. "And not only do we hear from patients all the time that they want to connect with other patients, but we know that researchers and industry also benefit from learning more about the patient experience because it gives them context and perspective on their work. Our LCA Family Conference accomplished that ten-fold this year."

Family members and advocates from throughout the country and Mexico said they appreciated the depth and quality of information presented at the conference that included information from other rare disease groups, the Young Adult Sickle Cell Alliance and the Barth Syndrome Foundation, on how they approach patient advocacy and patient life.

Joy Goodwine of upstate New York, whose 4-year-old daughter Jordynn has LCA2 (RPE65), 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."

Learn more about the conference presentation at sofiasees.org/resources/blog/

Kids with LCA made new friends!

Sofia Sees Hope staff were on hand for the conference

Annette Tonti, executive director of Sofia Sees Hope, moderated a panel called "Your Voice Matters!"

Kids had a field trip to LEGOLAND® Discovery Center

CRISPR/Cas9 Clinical Trial Recruitment for CEP290 Highlights Progress in LCA Research

By Ben Shaberman
Senior Director, Scientific Outreach and Community Engagement
Foundation Fighting Blindness

The landscape for Leber congenital amaurosis research continues to expand impressively with more emerging therapies moving into or toward clinical trials. Here is an update on many of those research efforts:

Editas Medicine and Allergan recruiting LCA10 patients for CRISPR/Cas9 study

Allergan, a global pharmaceutical company, and Editas Medicine, a developer of gene-editing therapies, have begun patient recruitment for a Phase 1/2 clinical trial for a CRISPR/Cas9 treatment for people with LCA10. The treatment targets a specific mutation (c.2991+1655A>G in Intron 26) of the gene CEP290.

Known as the Brilliance clinical trial, the study is the first for a CRISPR/Cas9 treatment for an inherited retinal disease. It is also the first clinical trial for a CRISPR/Cas9 therapy administered inside the human body. Brilliance is a dose-escalation trial that will enroll adult and pediatric (3–17 years of age) patients at sites in the United States.

The CRISPR/Cas9 gene-editing technology developed by Allergan and Editas is designed to locate and remove the mutation in LCA10. The treatment works like a pair of molecular scissors to cut out the mutation, and it is delivered to photoreceptors by a subretinal injection.

Gene editing is different from gene (replacement) therapy. In gene therapy, copies of an entirely new gene are delivered to the retina to replace the defective copies. In CRISPR/Cas9 gene editing, only the mutated region of the gene is corrected.

First Patient Receives AON Therapy for LCA10 in ProQR's Phase 2/3 Clinical Trial

ProQR, an RNA therapy development company in the Netherlands, has dosed the first patient in its Phase 2/3 ILLUMINATE clinical trial for sepofarsen. The treatment, formerly known as QR-110, is designed for people with LCA10 caused by the mutation p.Cys998X in the gene CEP290. Sepofarsen is an antisense oligonucleotide (AON), which works like "genetic tape" to block the mutation.

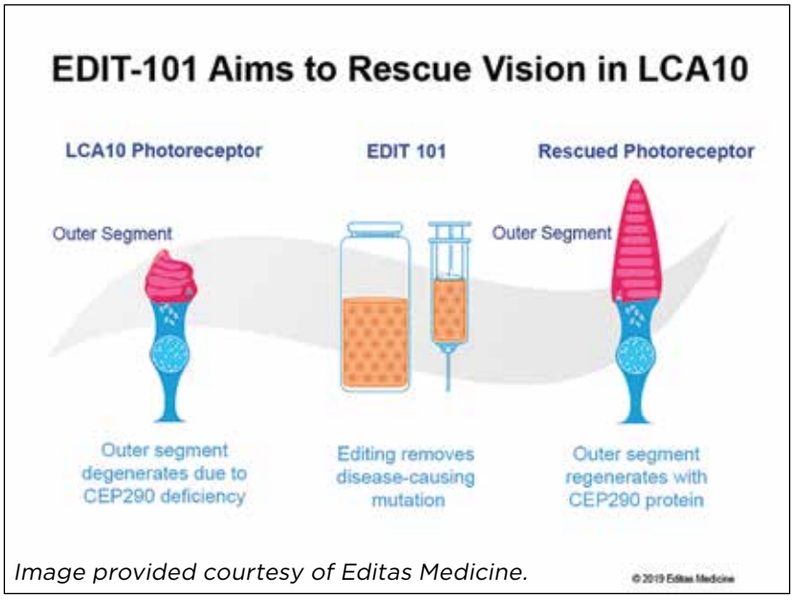

Image provided courtesy of Editas Medicine.

The 24-month trial will initially enroll 30 adults and children. Participants will be randomly assigned to one of three groups: 10 patients receiving 40 micrograms of sepofarsen, 10 receiving 80 micrograms of sepofarsen, or 10 receiving a sham procedure (placebo). The treatment is delivered by an intravitreal injection. The second dose will be administered three months after the first. Subsequent doses are administered every six months.

New LCA8 (CRB1) research at Duke University

Jeremy Kay, Ph.D., at Duke has received a three-year, $300,000 grant to identify the optimal CRB1 protein for treating people with mutations in the CRB1 protein, which can cause both LCA8 and retinitis pigmentosa (RP12). The CRB1 gene can express different forms (isoforms) of protein. Dr. Kay believes he has identified a CRB1 isoform that will work well in a gene therapy for people. He is evaluating the rescue efficacy and expression pattern of this isoform. His efforts will help CRB1 gene therapy developers design the optimal gene therapy for people with CRB1 mutations.

The Foundation Fighting Blindness is also funding Jan Wijnholds, Ph.D., at Leiden University Medical Center in the Netherlands, for development of a gene therapy for people with CRB1 mutations.

LCA1 (GUCY2D) gene therapy advancing toward clinical trial

Shannon Boye, Ph.D., at the University of Florida, is partnering with Sanofi to advance a gene therapy for LCA caused by GUCY2D mutations into a clinical trial. Earlier research for the project was funded by the Foundation. Dr. Boye hopes to begin the trial in 2019.

LCA4 (AIPL1) gene therapy available through compassionate use program

MeiraGTx, a gene therapy developer in the United Kingdom, has a compassionate use program for its AIPL1 gene therapy. A compassionate use therapy is an unapproved treatment that is made available to patients with serious conditions. The company is also conducting a gene therapy clinical trial for LCA2 and retinitis pigmentosa (RPE65 mutations).

Partnership formed for development of LCA6 (RPGRIP1) gene therapy

PTC Therapeutics, the nonprofit Odylia Therapeutics, and Mass Eye and Ear are collaborating on the development of a gene therapy for LCA6, which is caused by mutations in the gene RPGRIP1. The Foundation funded earlier research for RPGRIP1 at Mass Eye and Ear.

Limelight Bio developing LCA13 (RDH12) and LCA5 (lebercillin) gene therapies

Under the scientific leadership of Jean Bennett, M.D., Ph.D., Limelight Bio is conducting lab research for the development of gene therapies for LCA13 (RDH12) and LCA5 (lebercillin). The company's goal is to move these emerging treatments into clinical trials.

For more information on clinical trials, visit www.clinicaltrials.gov. More information on clinical and lab research is at available at www.fightingblindness.org.

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

Navigating Adulthood by Jack McCormick

Some Insight Into Getting a Guide Dog

Jack and his guide dog, Jake

When people ask me what it is like traveling with a guide dog, I tell them that it's like traveling with a GPS. With a map, you plan out your route and decide how you will get somewhere, but with a GPS, you tell it your final destination, and it determines how you are going to get there. This is like traveling with a guide dog because you don't need to remember landmarks in the same way. After you go somewhere once, the dog remembers it, much like saving a location in your favorites on a GPS.

The ways a guide dog changes how you travel is one of the many things to consider when deciding to get a guide dog. It is a big decision to make, and I am going to share my experience in the hope of making the decision easier for you.

I decided to get my guide dog Jake at the perfect time in my life—right before moving to a large city for university. I knew that the busy campus would be hard to navigate and my guide dog certainly made it easier—he remembered where all my classes were; would find me empty seats in lecture halls; and found routes through busy crowds for me.

There were also a number of benefits to getting a guide dog that I didn't think of before getting him—I stopped bumping into things (no more constant bruising on my shins); I was able to move much faster; and pretty girls stopped to talk to me about my dog, although more times than not they were more interested in him than me.

That being said, getting a guide dog is not all sunshine and rainbows. Last I checked brown wasn't a color in the rainbow, but I've had to deal with lots of the brown stuff since getting Jake. That's right, they poop, and you have to clean it up! Owning a dog is a lot of work! For example, I like to sleep in on the weekends, but Jake still likes to be fed at 6:30 AM. You will also need to consider how you are going to pay for the dog. Food is around $60/month, and vet bills are expensive—this past Christmas Jake got a bladder infection, and the emergency vet bill cost me almost $500.

I could talk about getting a guide dog all day, but I only have so much room, so I hope that I've given you some things to think about before you decide if a guide dog is right for you.

*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***

When Self-Advocacy Creates Change That Benefits Everyone

By Rosanne Smyle

Kristen Steele is a trailblazer.

The 22-year-old paved a smoother road for those without vision by changing national massage-therapy exam protocol, writing policies and procedures, and proving herself as a competent, independent contractor who makes house calls to the elderly and the ill in rural Nebraska.

Kristen, who lives in Council Bluffs, Iowa, near the Nebraska line, forges through life with passionate determination. She finds the support she needs to be her best and passes her knowledge along so others can be their best.

Doctors diagnosed Kristen with Leber congenital amaurosis as an infant, although her clinical diagnosis of LCA-CEP290, also known as LCA10, came in middle school.

She began reading Braille at age 3, and since first grade, has used a BrailleNote, a mini tablet-sized personal digital assistant with input through a Braille keyboard.

Struggling in school and having trouble with teachers adapting to a blind student, Kristen's mother quit her job to help her daughter. Her mom would pull up her assignments on the computer and tell her what she was missing and what she needed to go forward with the work.

"I didn't let anything stop me," Kristen said. "Sometimes I'd be up 'til midnight doing papers. I spent countless hours. I had to do it because I couldn't let myself fall behind because of someone's ill-preparedness."

Kristen graduated ahead of her class in December 2014, with plans to become a high school English teacher. That changed when she met a blind English teacher in Indiana.

"She told me to think twice about it" because she would often work from 6:00 AM to midnight and the job took over her life.

Kristen and Corvette after a shopping trip

CHOOSING HER PATH AND FIGHTING HER FIGHT

Kristen decided her passions aligned more with the medical field, given her interest in the healing arts. Also, her grandmother, who had dementia, had recently died, and she thought more about geriatrics and helping the elderly.

She reached out to a blind friend who is a licensed massage therapist and researched massage therapy schools. She ultimately enrolled at Midwest School of Massage near Omaha. The school turned out to be a perfect fit.

After completing the 1,000-hour course in February 2017 on anatomy, physiology, and pathology, plus 200 practice massages, with a 4.0-grade point average, she studied for the Massage and Bodywork Licensing Examination, known as the MBLEx.

Before taking the exam, Kristen took on the Federation of State Massage Therapy Boards because it would not administer the test in Braille. She would have to take the four-hour test with a hundred multiple-choice questions using a human reader, a volunteer likely unfamiliar with medical terms.

Through the National Federation for the Blind, Kristen found a blind lawyer in Iowa and they sued the agency. Eight months later, they reached a settlement agreement, making Kristen the first to advocate for and pass the MBLEx in Braille.

"I wanted to create this advocacy for anyone else pursuing massage therapy and let the boards know it should be in Braille."

FIRST STEPS DOWN A BUMPY CAREER PATH

Massage therapists often work as independent contractors, and Kristen's first contracted job led her to a brand-new company.

"They were accepting and very welcoming at first," she said. In fact, she helped write the company's new policies and procedures for its massage therapy program.

But her employer tried to have her sign a contract that she couldn't read because it wasn't in Braille. Her mother came with her to read the new contract, but the company initially did not produce it.

"They finally spilled it. 'We need to lower your rates.' "

She brought the case to the Nebraska Department of Labor and told officials she never looked over the contract because she can't see. She won.

Kristen applied for another job, but "They just didn't believe in me, that I would be able to find transportation, find the patients. They didn't have the confidence that the blind could do it."

She worked at a physical therapy clinic but left because of few hours and low pay. Then she found a program called "Comfort Touch: Massage for the Elderly and the Ill."

Comfort Touch™ instructor and licensed massage therapist Mary Kathleen Rose never had a Braille reader, so her course materials were not available in Braille.

Kristen ordered a print copy through Bookshare.org, an online library for people with visual disabilities. Bookshare scanned the book on a Monday and uploaded it by Friday, and she began the class.

"I was the first one to put these materials into Braille…and it was really cool because Mary Rose did a video of me reading in Braille the Comfort Touch™ textbook. I had adapted these course materials and paved the way, and for my turn (on the video) she wanted me to read 'Adapting to Change,'" which deals with loss, aging, and change.

EXPANDING HER HORIZONS

Kristen also earned certificates in hot-and-cold stone therapy, aromatherapy, Reflexology, advanced dementia processes, and she is a licensed massage therapist in Iowa and Nebraska.

She familiarized herself with Aira and Seeing IA, visual interpreter services. She sent her resumé to Aira and worked with a professional who formatted and polished her draft into what Kristen called the perfect resumé.

"It placed my disability on the back burner, and it gave me the upper hand when you have sighted massage therapists, and they're interviewing without any of these advanced certificates."

Between her resumé and an initial phone interview, Kristen felt she would be judged equally as a sighted person before showing up for an in-person interview.

After seeing a familiar job posting in May 2018, she sent her resumé to the company where representatives a year earlier did not believe she could do the job. She received a call back in an hour and did a phone interview.

During her in-person interview, Kristen demonstrated Aira—using a phone and wearing glasses connected via Bluetooth to a hotspot—and called an agent, displaying her ability to navigate a client in-take process and get around the office. Or, as she said, "I took Aira for a spin. I walked around the office, read people's name tags, saw suite numbers."

The company hired her the next day and she's currently thriving there as a massage therapist specializing in geriatric care.

Kristen proudly shows off her Massage Therapy certificate

Kristen, her dad and Corvette

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 • RARE in the Square

January 13–15, 2020 • San Francisco, CA • globalgenes.org/event/rare-in-the-square/

Set in San Francisco's Union Square, RARE in the Square brings together rare disease innovators to forge partnerships and advance innovation.

World Orphan Drug Congress USA 2020

April 29–May 1, 2020 • Oxon Hill, MD

terrapinn.com/conference/world-orphan-drug-congress-usa/index.stm

World Orphan Drug Congress USA presents its Global Orphan Drug Conference and Expo featuring 1,500 leaders in orphan drugs from 50 countries. Subjects include clinical development and manufacturing of cell and gene therapies, artificial intelligence and digital health solutions.

Foundation Fighting Blindness
VISIONS 2020 National Conference

June 18, 2020 • Minneapolis, MN • fightingblindness.org/events/visions-2020

VISIONS is the only event of its kind—created solely for individuals and families affected by retinal diseases. Find access to the latest retinal research and clinical trials, the doctors performing the work, and families from around the country living with the same diseases.

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

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