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

# SEEING HOPE | Newsletter

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May 2020 | Issue 8

## Kody Keplinger: Living Life as a "Confident and Proud Disabled Woman"

***By Rosanne Smyle***

Kody Keplinger, diagnosed at age 8 with Leber congenital amaurosis (LCA), just published her ninth young adult novel. *Lila and Hadley* tells the story of Hadley, an angry 12-year-old navigating life with degenerative vision because of retinitis pigmentosa (RP), and Lila, the abandoned dog she's helping prepare for adoption.

Kody—28 and living in New York City with her beloved guide dog, a German shepherd named Corey—grew up in rural western Kentucky and came to the city via Ithaca College. She characterizes herself as "a confident and proud disabled woman."

*The New York Times* best-selling young adult author has received a host of Young Adult Library Services Association awards. She won her first two accolades for *The DUFF* (Designated Ugly Fat Friend), summarized as: Could enemies-with-benefits become friends? She was still in high school when she wrote the book, which was made into a major motion picture in 2015.

Diagnosed legally blind as a toddler and later with LCA, Kody said in an article discussing the rare retinal disease: "LCA, as it's called, has many variants on many different genes, several of which (like mine) haven't yet been discovered."

More than 25 forms of LCA are known to exist. Kody's LCA leaves her with poor light perception and tunnel vision.

Kody writes in her message to readers of *Lila and Hadley* that she knew she wanted to write about a girl and a dog, that she'd had dogs all her life and that it seemed odd she hadn't previously written about that bond.

***Continued on page 3***

## Sofia Sees Hope Gives More Than $100K in 2019 For Research and Genetic Testing for Inherited Retinal Disease Patients

***By Elissa Bass***

Sofia Sees Hope donated more than $100,000 at the end of 2019 to support research into treatments for inherited retinal diseases (IRDs), including Leber congenital amaurosis (LCA), and to provide free access to genetic testing for those with IRDs.

Since its founding in 2014, Sofia Sees Hope (SSH) has directed $255,000 to research and given $125,000 to provide families free access to genetic testing. This is in addition to providing outreach support, education and advocacy to the IRD and LCA patient communities through programming and a biennial family conference.

***Continued on page 2***

# Sofia Sees Hope Gives More Than $100K in 2019 For Research and Genetic Testing for Inherited Retinal Disease Patients

*Continued from page 1*

"We are pleased to once again be able to support these researchers and their work to find treatments for LCA," said SSH co-founder Laura Manfre. "With nearly 30 different genetic variations causing LCA, it is equally important to us to provide access to genetic testing for families who are in the early phases of their diagnostic journey. Having spent seven years looking for a genetic diagnosis for our daughter Sofia, we know how expensive and overwhelming that journey can be. Anything we can do to alleviate road blocks for people in the IRD community is important to us."

**The 2019 donation to Foundation Fighting Blindness includes:**

- **$15,000** to Sandro Banfi, Fondazione Telethon, Italy: AAV-Sponge-mediated modulation of microRNA-181a/b: a potential therapeutic approach for Inherited Retinal Disease
- **$20,000** to Clay Smith, University of Florida: Enhancing Metabolism in Photoreceptors with a Modified Arrestin to Treat Retinal Degeneration
- **$25,000** to Krishanu Saha, University of Wisconsin-Madison: Gene Editing Nanomedicines to Correct Pathogenic Mutations in the Retina
- **$25,000** to Rob Collin, Radboud University Medical Center, the Netherlands: Development and optimization of AON-based therapies for selected splice defects

Sofia Sees Hope has supported Drs. Saha and Collin in the past with research funding. Dr. Collin's earlier work that was funded in part by SSH is now with the Dutch biotech, ProQR, which is developing a novel drop, sepofarsen (QR-110), for patients with LCA10 due to the p.Cys998X mutation, also known as c.2991+1655A>G mutation, in the CEP290 gene. Top-line results from a recently completed Phase 1/2 clinical trial in children and adults suggest that the majority of patients treated with sepofarsen experienced a substantive overall improvement in vision at 12 months of treatment.

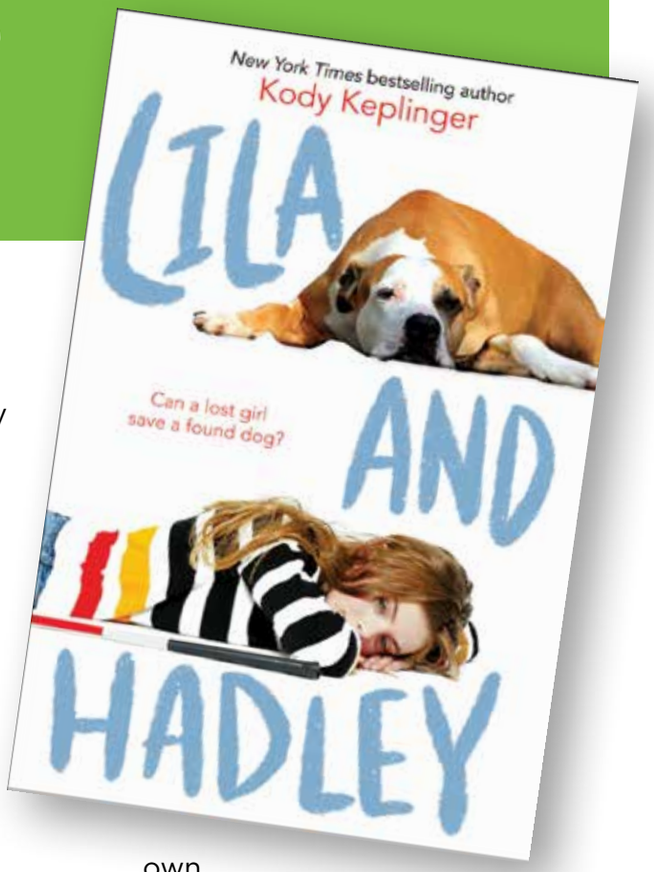
Additionally, Sofia Sees Hope donated $23,200 to My Retina Tracker to provide free access to genetic testing. The My Retina Tracker Registry is a research database of people and families affected by rare inherited retinal degenerative diseases (IRDs). It is designed to share de-identified information within the IRD research and clinical communities about people with an inherited retinal disease to help accelerate the discovery of treatments and cures. SSH made an initial $15,000 donation to the program in December, and was able to add $8,200 in Q1 of 2020 from its 2019 year-end appeal, which pledged every dollar raised to benefit free genetic testing.

"The support that Sofia Sees Hope has given helps ensure that the most promising research will advance to clinical trials and that My Retina Tracker Registry will continue to register thousands of affected individuals who may be helped to regain their vision or slow its loss by enrolling," said Benjamin Yerxa, PhD, chief executive officer at Foundation Fighting Blindness.

Sofia Sees Hope is able to support research and provide access to genetic testing because of its successful fundraising, primarily through its signature gala event, Dinner in the Dark. Matching corporate gifts and other event fundraising proceeds make these donations possible, as well as a national conference for LCA families, and other outreach events throughout the calendar year.

# Kody Keplinger: Living Life as a "Confident and Proud Disabled Woman"

*Continued from page 1*




"But Hadley was one of the most challenging characters I've written, in large part, because we have something really personal in common," Kody tells her readers. "Hadley, like me, is legally blind. Her condition (retinitis pigmentosa) is very similar to my own in how it affects the eyes, but with one key difference: I was born legally blind, whereas Hadley's vision has been steadily worsening for years…"

Scholastic said in a news release: "*Lila and Hadley* serves to illuminate the sadness, anger and fear that can come with the challenges of disabilities in the hope for a more inclusive tomorrow. Hadley is angry about a lot of things: Her mom going to jail. Having to move to another state to live with her older sister, Beth, even though they haven't spoken in five years. Leaving her friends and her school behind. And going blind. But then Hadley meets Lila."

*Kirkus Reviews* had this to say about the book intended for readers ages 8 to 12: "A former Kentuckian and legally blind, Keplinger knows this territory, literal and figurative, inside out. A funny, moving tale, at once unsparingly realistic and upbeat."

Kody also has received high praise for her previous novels on topics that include a mass school shooting, ethics, relationships, social issues and self-awareness.

In *Run* (2016), best friends Bo and Agnes run away together to escape family stress. Agnes is blind from Leber congenital amaurosis. *Shut Out* (2011) plays out like a contemporary *Lysistrata*, in which the Hamilton High School girls, tired of their boyfriends' sports rivalry, won't see any action from them until they end their pranks.

*A Midsummer's Nightmare* (2012) has Whitley's father marrying the mother of a boy she hooked up with at a graduation party, while *That's Not What Happened* (2018) tells the story of Leanne (Lee), one of a handful of students who survived a school shooting, who needs to decide what to do when another victim's parents prepare to write a book with a false story. For more on her books, please go to Kodykeplinger.com.

Kody figured out early that she liked to write, beginning writing and drawing at age 2. Along with writing young adult books, she teaches writing at Gotham Writer's Workshops in New York City and she co-founded Disability in Kidlit, an online platform discussing the portrayal of disability in middle grade (ages 8–12) and young adult (ages 12–18) literature.

The platform's goals include helping writers create more authentic, accurate and respectful disabled characters. Disability in Kidlit's philosophy? "We believe that a thoughtful portrayal of disability requires more than memorizing a list of symptoms; we hope that sharing disabled people's thoughts on stereotypes, pet peeves, particular portrayals, and their own day-to-day experiences will help our readers learn the realities of disability, which are often different from what we see in popular media."

*Lila and Hadley* fills that bill because the story has broad appeal, as Kody said: "Everyone who's ever been frustrated or felt angry or felt alone or even just everyone who's ever had a pet that they've really, really loved, I think that there's a little bit of something here for every kid to find and I hope that readers will pick the story up and will fall in love with Lila the way Hadley does."

So do we, Kody.

***We hope in the future to bring Kody Keplinger to Sofia Sees Hope's "A Rare Opportunity" event. With limitations on gatherings because of the coronavirus, we canceled the planned April forum that included a talk and book signing by the author, along with visits to middle-school students. We'll keep you posted.***

# LCA10 Patient Receives First Emerging CRISPR/Cas9 Therapy in Clinical Trial

## *Is a clinical trial right for you?*


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

It is an exciting time for people with Leber congenital amaurosis (LCA) and other inherited retinal diseases (IRDs) thanks to dozens of clinical trials underway for potential therapies to save and restore vision.

Adding to the excitement are news stories from major media outlets, highlighting elegant, cutting-edge treatment-approaches that are now being evaluated in humans. For example, on March 4, National Public Radio (NPR) ran a feature story on the first time that an emerging CRISPR/Cas9 (gene-editing) therapy was administered inside the human body. The Phase 1/2 study at Casey Eye Institute, Oregon Health & Science University, happens to be for LCA10 (CEP290) patients.

Sponsored by Allergan, a global pharmaceutical company, and Editas Medicine, a gene-editing therapy company, the dose-escalation BRILLIANCE clinical trial will be assessing safety and efficacy of their emerging gene-editing therapy in 18 patients at four U.S. sites.

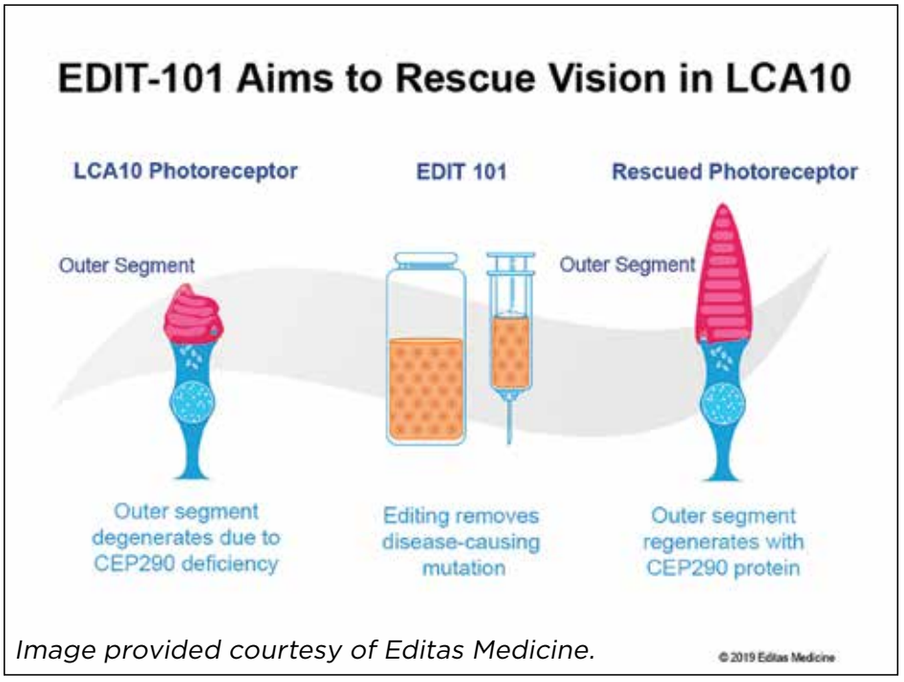



*Image provided courtesy of Editas Medicine.*

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.

The emerging treatment from Allergan and Editas Medicine targets a specific mutation (c.2991+1655A>G in Intron 26) of the gene CEP290, which, when mutated, causes LCA10. Known as EDIT-101, the CRISPR/Cas9 gene-editing technology is designed to locate and remove the above-mentioned mutation in CEP290. The treatment works like a pair of molecular scissors to cut out the mutation.

"This is an exciting milestone for using CRISPR/Cas9 gene-editing to potentially treat inherited retinal diseases," said Brian Mansfield, PhD, executive vice president and interim chief scientific officer at the Foundation. "Gene-editing is an attractive approach for addressing large genes that exceed the cargo capacity of commonly used viral delivery systems such as adeno-associated viruses or AAVs."

Sounds pretty cool, huh? Perhaps you or your loved one want to enroll in a study like this. You may be thinking: Finally, there may be something to save or restore my vision.

**But is a clinical trial right for you? There are many factors to consider:**

## CLINICAL TRIAL PARTICIPANTS ARE PIONEERS

Keep in mind that a clinical trial is research to determine if a POTENTIAL treatment is safe and effective. The molecule, gene/protein or cell being evaluated only becomes a treatment after the trial is completed and the treatment is approved by the regulators. A trial participant should not expect to benefit from the treatment. He or she might benefit, but the trial's purpose is to see if the therapy does, in fact, work. For the regulators (e.g., U.S. Food and Drug Administration), safety is first and foremost, and safety is the primary focus of the early phases of a clinical trial.

Perhaps most important for trial participants to know is they are pioneers helping to advance the research. Thanks to their commitment to the study, and regardless of whether it works for them, they are helping to advance the emerging treatment, potentially making it available to others.

## PARTICIPATION IS A BIG COMMITMENT

For those who do enter clinical trials, they are exposed to some risk of adverse events, though prior lab studies are intended to minimize that risk. Also, a participant usually will have to make many visits to the clinical center for evaluation and follow-up. The visits themselves can involve lots of tests. Trial sponsors (therapy developers) normally pay for any travel and accommodations. It is important for participants to keep their commitment to the trial, so that it can be completed. Participants dropping out can put the trial—and the future of the emerging therapy—in jeopardy.

## INCLUSION AND EXCLUSION CRITERIA—WHO QUALIFIES?

When researchers recruit for a clinical trial, at least in the later stages, they are looking for the patients who are most likely to respond well to the emerging treatment. For example, in many retina-related trials, people with conditions such as glaucoma and cataracts are usually excluded, because those conditions can impact vision independent of what's going on in the retina. Women who are pregnant (or not on birth control) may be excluded to protect an unborn child. People with other significant conditions (e.g., cancer) may not be allowed to enter the trial.

## GO TO CLINICALTRIALS.GOV

Every clinical trial in the United States, and many in other countries, are listed at www.clinicaltrials.gov, hosted by the National Institutes of Health. The database includes extensive information on each study, such as inclusion and exclusion criteria, sites for the trial, and contact information for the trial coordinators

***One word of caution:*** *Just because a trial is listed on clinicaltrials.gov, doesn't mean it is an FDA-authorized trial. If the trial sponsor or clinic is charging for the "emerging therapy," that is a major indication the trial is not monitored by a regulatory agency and could be unsafe.*

The Foundation Fighting Blindness currently funds the following CRISPR/Cas9 lab research projects:

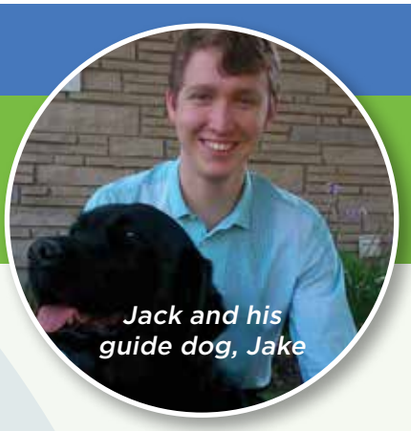
- University of Wisconsin-Madison (LCA16 caused by a mutation in KCNJ13)
- Mass Eye and Ear (Retinitis pigmentosa caused by a mutation in RP1)
- Mass Eye and Ear (Retinal disease caused by a mutation in USH2A)
- UCLA (Usher syndrome 1B caused by a mutation in MYO7A)

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

**The Foundation Fighting Blindness VISIONS 2020 Conference is now a virtual experience. FFB is actively developing a virtual experience that will provide a wide range of science and research presentations, practical adapting and thriving sessions and an opportunity to connect with others from across the country. Visit www.fightingblindness.org/visions-2020 for additional information as it becomes available.**

*Navigating Adulthood by Jack McCormick*

# Job Hunting: Research, Ask, Stay Positive


*Jack and his guide dog, Jake*

Unfortunately, in the vision-loss community you do not have to go far to hear horror stories from the workplace. Stories of interviewers asking candidates how many fingers they are holding up or companies terminating long-term employees due to declining vision are all too common. It's easy to get discouraged hearing these stories, however, from my experience there are things you can do to make sure that you are competing for jobs on an even playing field.

## DO YOUR RESEARCH

It is important to learn as much as you can about a job before you invest time and money to gain the qualifications to do that job. What are the career outlooks? Are the jobs in accessible locations? Is the technology that is used accessible? Will you enjoy the work?


I am a Human Resources (HR) professional. When I decided to enter this field, I learned that HR professionals are in demand and that most jobs are in accessible larger cities. I also learned that I would probably enjoy the work by speaking with people in the field. I do, however, regret not asking about the technology used in HR. Unfortunately, many small- and medium-sized companies still keep paper employee records, which is an accessibility barrier for me. That being said, I have been able to find great jobs, and good employers will find accommodations for these barriers. The point is, the more you know the better you can prepare.

## ASK FOR HELP

Everyone's accommodation needs are different, and you can't expect your employer to read your mind. It's important to ask for accommodations when you need them. It can be intimidating but remember that your employer is paying you a lot of money and they want you to be as productive as possible. If spending $800 on a new computer program will help you do your job faster, then it makes good business sense to spend that money.

You can also ask for accommodations in an interview. I currently work at a hospital. When I was invited for an interview I asked someone to meet me at the front door because I knew that it would be challenging to find the meeting room. Yes, I was scared they weren't going to be receptive but, I decided that if the employer wasn't willing to do something so simple for me at the interview stage, then it wouldn't be a good place to work anyway.

## DON'T GET DISCOURAGED

Applying for jobs is like dating—it's a numbers game and it takes a while to find the right one. The average job has 200+ people apply for it and only a few get invited for the interview. Keep applying and if you know someone who works for the company, let them know you've applied—they may be able to help land you that interview.

*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***

# Sofia Sees Hope Formalizes, Relaunches Ambassador Program

*Tami and Mike Morehouse — SSH Ambassadors*

***By Elissa Bass***

This spring, Sofia Sees Hope formally relaunched its Ambassador program, which has been operating for about two years.

The Ambassador program designates individuals within the rare inherited retinal disease (IRD) and Leber congenital amaurosis (LCA) communities around the United States and the globe as official representatives of Sofia Sees Hope. The goal is to provide more coordinated outreach and messaging on behalf of the nonprofit.

The Ambassadors—all of whom have been involved with the organization for some time—will focus on four keys areas for Sofia Sees Hope:

- **FAMILY OUTREACH AND SUPPORT:** Connecting LCA and IRD families with critical resources, support, and one another
- **EDUCATION AND RESEARCH:** Engaging with families, clinical and research specialists, and advocacy groups to share information, expanding the science and understanding of LCA and the diverse needs of the IRD community.
- **FUNDRAISING AND DEVELOPMENT:** Raising transformational financial support for SSH and LCA through the organization and marketing of fundraising opportunities and sponsorship.
- **ADVOCACY:** Building relationships with stakeholders and legislators at the local, state, and federal level to educate and inform policies related to LCA and IRDs.

"We've wanted to expand and add structure to this program for some time, as Sofia Sees Hope has grown," said nonprofit co-founder and board President Laura Manfre. "We are physically based in Connecticut, but our reach and our impact are global. Our Ambassador volunteers will allow us to expand our presence in this country and elsewhere. A vital element of the Sofia Sees Hope mission is to provide more opportunities for all people affected by LCA to connect and provide mutual support and information."

Ashley Luppold, an experienced and strategic learning and development professional, has been named director of the Ambassador program. "I am incredibly excited to support the Sofia Sees Hope Ambassador program," she said. "The passion and commitment within the LCA and IRD community is a powerful force. This program enables us to formalize and direct this power to extend the network of support, education, research, and advocacy for LCA/IRD patients and their families."

**This spring, Foundation Fighting Blindness is unveiling a fun, creative way to celebrate together with a virtual VisionWalk experience on June 6th.**

**How can you support our National Virtual VisionWalk?**
**Visit give.fightingblindness.org/VirtualVisionWalk to register your team and take advantage of our online tools.**
**Questions? Contact Michele DiVincenzo mdivincenzo@fightingblindness.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.*

**EDITOR'S NOTE:** *Please check each event's website for confirmation, as of publication these events were still scheduled.*

## Global Genes 2020 RARE Patient Advocacy Summit

**September 21–23, 2020**
**San Diego, CA**
**globalgenes.org/event/patient-summit**

As the largest gathering of rare disease patients, caregivers, thought leaders and other rare disease stakeholders in the world, the RARE Patient Advocacy Summit is an unparalleled opportunity to forge meaningful connections with other rare advocates and bring home actionable strategies and tools to accelerate change. Register now.

## National Organization for Rare Disorders (NORD) Rare Diseases and Orphan Products Breakthrough Summit

**October 8–9, 2020**
**Washington, D.C.**
**rarediseases.org/summit-overview**

This summit offers the opportunity to collaborate with more than 900 top leaders from the Food and Drug Administration, National Institutes of Health, industry, patient groups, payers and research institutions to address the progress and innovations in rare disease diagnosis, treatment, patient engagement and market access of orphan products. Register now for the event featuring more than 20 FDA speakers, more than 100 patient organizations, and the pharma/biotech industry's foremost experts in orphan product innovation, investment and commercialization.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**To learn more about Sofia Sees Hope, visit our website at www.sofiaseeshope.org.**

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