

SEEING HOPE Newsletter

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From the Founder:

As 2023 comes to a close, we have much to celebrate at Hope in Focus and many thanks to give to you.



Laura Manfre

Our 8th Dinner in the Dark fundraiser came back stronger than ever this year. We are filled with gratitude and appreciation, as we are happy to announce we raised more than \$200,000 to help drive our mission of transforming the lives of those affected by blindness by Leber congenital amaurosis and other rare inherited retinal diseases (IRDs).

We also celebrate the many connections made this summer among LCA families and friends at our 3rd Hope in Focus LCA Family Conference in Indianapolis. Our Family Conference has become *the* go-to event for individuals living with LCA.

Hope in Focus also made an important connection with the Food and Drug Administration this fall by hosting an FDA Patient Listening Session, in which people living with LCA and their caregivers shared with agency staff their heartfelt experiences and urgent pleas for treatment.

We look to 2024 with renewed efforts to raise funds to advance

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Hope in Focus Brings Voices of People Living with LCA Directly to the FDA

By Paul H. Hebner

Allison Wolf, whose 9-year-old son Elliot lives with the encroaching blindness of Leber congenital amaurosis (LCA), spoke directly to staff from the country's regulatory agency to help them understand the dire need for treatments for this rare inherited retinal disease.

"Elliot's eyes are dying," Allison said. "They are dying and we have nothing to help slow down the progressive loss of his eyesight," she told the Food and Drug Administration (FDA).

"What would a timely and effective treatment mean? The whole world," the mother of four said.

Allison and five others affected by LCA shared their stories with the FDA during a recent Patient Listening Session hosted by Hope in Focus.

We brought together the voices of people living with LCA, caregivers, and a clinician treating people with LCA to FDA



Elliot Wolf

staff members in a 90-minute online meeting Oct. 30, 2023. Simply put, we wanted the FDA to truly hear our community's voices.

Our year-long planning resulted in a successful session to help regulators understand patient/caregiver experiences related to LCA. We requested the meeting to raise awareness with FDA staff about clinical differences of LCA genotypes, share patient experiences of LCA's impact on the quality of their lives, given the scarcity of treatments, and communicate the extraordinary significance of vision stabilization for patients.

RAISING AWARENESS IS KEY TO ADVANCING RESEARCH

People in our community struggle daily to find ways to cope and try to live normal lives in a sighted world. Still,

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Hope in Focus Brings LCA Voices to FDA

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they never give up hope for new treatments and the possibility of a cure. As an advocacy organization, one of our biggest challenges is creating awareness of LCA.

Courtney Coates, our Director of Outreach and Development, introduced the Hope in Focus mission and said LCA affects about one in 33,000 people and presents early in life, resulting in total blindness. Twenty-seven genes have been identified to cause LCA, with treatment available for one form, while a handful of others are in clinical trials.

Co-founder and Board Chair Laura Manfre said as the parent and caregiver of their daughter Sofia living with LCA, raising awareness, searching out new treatments, and providing support to the growing LCA community became the mission of Hope in Focus.

Laura said she wanted to underscore the goals of treatment, in particular around the stabilization of vision for Sofia and others with LCA.

“While vision restoration would be fantastic, having access to a treatment that stabilizes her vision would be huge for her. Not having to constantly change her accommodations for her changing vision is a huge win, and not having to fall asleep every night worried that the next morning might be the one she wakes up without any vision at all. That would be life changing.”

Sharing a chart showing seven current clinical trials, she explained two are not recruiting at this time, two have been discontinued, and all seven represented only four LCA genetic variations.

“So, there is still a lot of work to be done to advance potential treatments.”

CAREGIVERS AND PEOPLE LIVING WITH LCA TELL THEIR STORIES

Six people from our community shared their experiences.

Allison Wolf, who is 45 and from North Dakota, said doctors diagnosed her son Elliot with LCA13 (*RDH12*) when he was 6, and as he gets older, her challenges also grow.

“Nobody knows how to raise a visually impaired child. I must be taught. I must teach him the sighted world, while also dealing with the challenge of teaching him the non-sighted world,” she told agency staff.

“My heart hurts when I hear Elliot say, ‘I just want to be done with all this work and I just want to play.’”



Elliot Wolf

Mohamed Farid of Illinois is a 36-year-old entrepreneur, diagnosed with LCA5 at 40 days old. His mother quit her job to take care of him, and they moved back to Egypt. Preserving his sight concerns him the most, and his greatest fear is the risk of injury.



Mohamed Farid

“For new treatments,” Mohamed told the FDA, “the first thing I would look for is something to stop any degradation — I sense that my functional vision has degraded slightly over time and would imagine there is something going on with my retina, but do not know. I have been told by doctors that I

will lose my sight in the next two to three years ever since I was 5, and that lack of a prognosis is a killer. Following conservation, I care most about night blindness, then field of vision. ...

“I was lucky to have a huge support network of people, who went out of their way and had a lot of energy and stress tolerance. This does not mean that life was easy or that I would not want a cure or treatment, even a partial cure would be life changing. It does mean that the cost of LCA is high, but barely bearable.”

Joy Goodwine, 40, of New York is the mother of 7-year-old Jordynn, who was diagnosed at age 1 with LCA2 (*RPE65*) and received a new diagnosis last year of LCA1 (*GUCY2D*). She always wanted Jordynn to feel a sense of normalcy.

“I’ve always asked family and friends to treat her like any other child,” Joy said. “I do not want people to feel sorry for her, and I do not want her to have any special treatment because of her visual impairment.”



Jordynn Erwin

Jordynn is being raised to know that she is capable of doing anything she wants with limitations. ...

“What I fear most as a parent with a child living with LCA is I worry that her kindness and vulnerability will lead to unwanted interactions when she gets old enough to go off on her own, or her eyes get worse if treatment is not available.”

Linda Wirth is 76 and lives in Colorado. Her vision loss remained a mystery until her diagnosis of LCA10 (*CEP290*) at age 68. As a child, she learned about the world around her by touching things and holding them.

"My family often told me, 'Don't touch that! You will break it. Don't act strange. People will stare at you.' So, I began to fake it, pretending I understood things which I did not in order to get along and not appear any different."



Linda Wirth

One doctor early on told her: "You are blind! What do you want me to tell you? If you are looking for a miracle, there is none."

A retired clinical social worker, Linda's been angrily dismissed from jury duty and job interviews, ignored by waitstaff, and questioned whether she deserved to be a parent.

"Just as many differently abled folks, I have encountered individuals with various stereotypes and prejudices."

DJ Broadbin of Connecticut is a 34-year-old mother and caregiver to 5-year-old Jace, who was diagnosed with LCA4 (AIPL1) at 10 months old. Through a Hope in Focus connection, she succeeded in getting compassionate use treatment in the UK for Jace. The treatment, which applied only to his specific gene mutation, has restored some of his sight in one eye.

"No matter what happens with his vision or developing new treatments," DJ told the FDA, "I remain steadfast in having the same



Jace Broadbin

goal I had for Jace since the very second I found out I was going to be a mom — I just want him to be happy. And I will continue to fight for his inclusion in this world to make sure that he always is."

She often finds herself exhausted by the constant stress of being a caregiver.

"I also wish sometimes," DJ said as her voice broke, "I just got to be a mom. Not a medical coordinator, an occupational therapist, and a Teacher of the Visually Impaired all rolled up into one. Just mom."

"But then I think about all the things that Jace's diagnosis has given me personally: Clarity. Patience. Compassion and empathy in a way that I myself was too blind to see in those earlier days."

Mirielle St. Arnaud is 16 and lives in Illinois. She was diagnosed with LCA IQCB1/NPHP5 at 6 months old. Her early diagnosis was possible because her older brother had been diagnosed with LCA. Mirielle worries about her safety and struggles to fit in with her classmates because she misses social cues. She hopes her fairly stable vision will not get worse.



Mirielle St. Arnaud

She shared a wish list with the FDA that included personal independence, the opportunity to participate in sports, and more certainty about achieving the career of her choice (she dreams of being a lawyer).

"Some of the more significant challenges I face because of visual impairment relate to jobs and getting hired. It takes longer for me to learn certain skills at a job. I have to rely on others when out in public, especially when traveling. I also struggle to find accommodations for my impairment. It seems that other people know my needs better than I do."

From the Founder:

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treatments for LCA and IRDs, as we head into our 10th year at Hope in Focus.

Thank you for being such an important part of our vital mission.

With gratitude and joy,

Laura

Our representative clinician, **Rachel M. Huckfeldt, MD, PhD**, of Massachusetts Eye & Ear, spoke about the difficulties of diagnosing and managing LCA and the small number of clinical trials. Identifying the responsible genes can be difficult, and the small number of patients severely limits opportunities for clinical trials.



Rachel M. Huckfeldt, MD, PhD

Questions from FDA staff members focused on a few common themes: Have you participated in clinical trials? What are the key impediments to clinical trial participation? What are your deal breakers to participation? What would a successful treatment look like to you?

One result certainly not on the agenda was the deep emotional impact of the stories shared by those living with LCA and caregivers. The session manifested itself as a profoundly moving event.

A full summary of the FDA Patient Listening Session is available on our website at hopeinfocus.org.

Hope in Focus Grateful for 2023 Dinner in the Dark Success



By Paul H. Hebner

So much more than a typical fundraiser, the 8th Hope in Focus Dinner in the Dark we advertised as “a unique sensory experience,” gave guests just that and much more Saturday, Oct. 14, 2023, at the Mystic Marriott Hotel & Spa in Groton, Connecticut.

Dinner in the Dark afforded the opportunity for guests to experience their dinner in a different way — wearing blindfolds. For a short time it offers a small taste of what it might be like for someone dealing with a rare retinal disease. This key fundraising event supports our mission of “Seeing a cure for blindness,” as we advocate for people living with Leber congenital amaurosis (LCA) and other rare inherited retinal diseases (IRDs).

Hope in Focus counted 273 attendees this year, with the event raising more than \$200,000, a record for our annual fundraiser. The evening is about bringing people together to connect with the mission of Hope in Focus, as we work to transform the lives of those

living with blindness caused by LCA and raise funds to support research treatments.

Laura Manfre, Hope in Focus Co-founder and Board Chair, said even as science advances, only one treatment is on the market for just one form of LCA.

“We need to do better. We need to continue advancing the science, but also move the science from the lab and the clinic to the community waiting for treatments.

“It is with deep gratitude,” she said, “that we thank all the people who came out to support our efforts to improve the lives of people living with rare retinal diseases and help advance research for treatments. We could not do this without you, and we are forever grateful for your contributions toward our mission.”

Courtney Coates, Director of Outreach and Development, thanked each of our sponsors and the Dinner in the Dark Committee members — without

whom the event would not have been possible — for volunteering their countless hours behind the scenes.

Laura introduced the evening’s key presenter, Dr. Daniel Chung, as a rock star. Dr. Chung is Chief Medical Officer for SparingVision, an ocular genomic medicine company. His work focuses on therapies to combat blinding diseases. He made a point of praising the courage of participants in clinical trials. These patients offer themselves to science, not knowing what the result may be, or if there will be any result at all.

“The real rock stars,” Dr. Chung said, “are those who say, ‘I’m going to be part of a clinical trial.’”

Sofia Priebe, the daughter of Laura and her husband, Chuck Priebe, spoke via a video presentation to offer tips for a successful dining experience. Hope in Focus began because Sofia was diagnosed with LCA at age 2.

Now a junior in college, Sofia spoke with the poise and grace of someone beyond her years. She advised on such techniques as determining which side of your knife is sharp, using utensils to explore the food on your plate, and remembering to always identify yourself and the person you're speaking to in conversation at the table.

In her most important tip, she said: "Food to face, not face to food."

Karen Clarke of Guilford said navigating a meal while blindfolded is challenging and humbling.

"The experience gives you a small glimpse into the challenges so many deal with every day. It's a great motivation for us to help with research — and be thankful for our own sight when the blindfold comes off."

Rosalie and John Maxham of Stonington attended the dinner for the first time.

"We came away appreciating this organization devoted to helping find a cure for a disease we never knew existed, and we were very happy to lend our support to this zebra."

We at Hope in Focus thank all the participants and faithful sponsors of Dinner in the Dark and look forward to an exciting event in 2024.

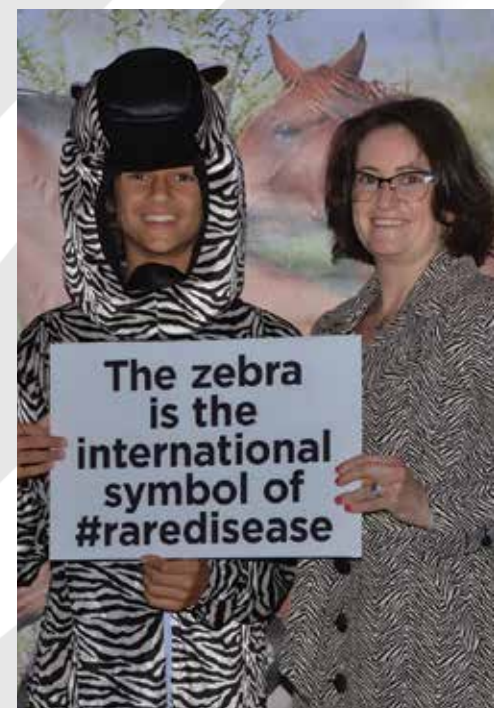
Please visit hopeinfocus.org for a complete list of our sponsors, and the menu, auction items, and photos.

Photos by: PubliCreatives



WHY STRIPES FOR RARE DISEASE?

Medical students are told to look first for the most common diagnosis. That's why they were taught, "When you hear hoof beats, think of horses, not zebras." Well, since we're raising funds to treat rare eye disease, we want you to think of zebras, the international symbol of rare disease. Thanks for showing off your stripes at Dinner in the Dark!



Update from the LCA Clinical Trial Front Lines



Ben Shaberman
*Vice President,
Science Communications
Foundation Fighting Blindness*



I am always excited when emerging therapies for inherited retinal diseases (IRDs) advance into human studies. The preclinical journey is long and painstaking. It can take five to 10 years (sometimes longer) to develop a therapy and move it out of lab studies into a clinical trial. Advancing a treatment across that threshold also takes millions of dollars, manufacturing expertise, and strong knowledge of the regulatory process.

I am pleased to provide updates from two early-stage clinical trials for promising therapies for Leber congenital amaurosis (LCA).

Opus Genetics launches clinical trial for LCA5 (lebercilin) gene therapy

Opus Genetics, a company developing gene therapies for people with IRDs, has dosed the first patients in its Phase 1/2 gene therapy clinical for LCA5, which causes significant vision loss in children. The Phase 1/2 clinical trial is being conducted at the University of Pennsylvania. Once safety in adults has been established and confirmed by the Food and Drug Administration, Opus plans to dose pediatric patients.

Known as OPGx-001, the gene therapy uses a human-engineered, adeno-associated virus to deliver healthy copies of the LCA5 gene to the retinas of patients, augmenting the mutated copies causing vision loss. The LCA5 gene expresses the lebercilin protein, which is important for the development and function of photoreceptors, the retinal cells that make vision possible.

OPGx-001 is administered through a one-time injection underneath the retina. Researchers believe gene therapies will be effective for many years, perhaps the life of the patient.

The LCA5 gene-therapy clinical trial is the first launched by Opus, a company conceived and formed by the Foundation Fighting Blindness. Founded in 2021, Opus received \$19 million in seed funding from the Foundation's Retinal Degeneration Fund, a venture philanthropy fund for emerging retinal disease therapies in, or nearing, early-stage clinical trials.

Ocugen recruiting LCA10 (CEP290) patients for its modifier gene therapy clinical trial

The biotech company Ocugen recently provided an update on the Phase 1/2 clinical trial for OCU400, its emerging, modifier gene therapy, which delivers copies of the *NR2E3* gene to improve regulation of multiple functions in the retina, including: photoreceptor maintenance and development, metabolism, phototransduction, inflammation, and cell survival. The company says OCU400 is designed to work for people with IRDs caused by a broad range of gene mutations.

The trial is currently enrolling adult and pediatric patients with LCA10 caused by *CEP290* mutations, as well as pediatric patients with rhodopsin (*RHO*) and *NR2E3* mutations.

Thus far, the trial has enrolled people with: retinitis pigmentosa (RP) caused by autosomal dominant mutations in *RHO*; RP caused by autosomal dominant mutations in *NR2E3*; RP, enhanced S-cone syndrome; and Goldmann-Favre syndrome caused by autosomal recessive mutations in *NR2E3*.

The latest report from the trial is for 12 patients who had follow-ups ranging from six to 12 months after subretinal injection of OCU400 in one eye. OCU400 had a favorable safety profile in the Phase 1/2 clinical trial.

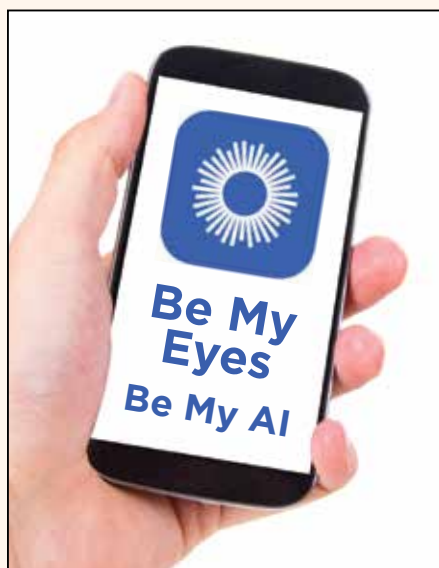
Also, eight of 12 patients had stabilization or improvement in all three of the following visual function measures: best corrected visual acuity, low luminance visual acuity, and navigation of a multi-luminance mobility test.

Visit **[FightBlindness.org](https://fightblindness.org)** to stay informed about the latest research advances for LCA and other IRDs.

AI — A Game Changer for People with Low Vision or No Vision

Artificial Intelligence — AI — is changing our world. Some people are nervous, unsure of the impact it will have on jobs, human creativity, and more. I'm excited by what I've seen and the potential it has to change my life!

While still in its infancy, AI can do a lot. *Be My Eyes*, a free app for receiving video support at a moment's notice, has been empowering blind and visually impaired people for years by connecting them with sighted volunteers who can help with visual tasks like checking the expiry on a product.



Be My Eyes recently introduced a new feature called *Be My AI* that I am currently beta testing. With *Be My AI*, I can have photos

described to me several seconds after taking them and ask the AI follow-up questions about it. For example, here is a description of a photo I just took of my kitchen:

"The picture shows a kitchen. The kitchen has dark brown cabinets with silver handles. The countertops are made of a speckled granite material. On the left side, there is a sink with a faucet and a small dish rack with a few dishes in it. Next to the sink, there is a toaster and a container with kitchen utensils like spatulas and spoons. On the right side, there is a stainless-steel stove and oven with a towel hanging on the handle. Above the stove, there is a microwave. The kitchen also has a ceiling light and an air vent on the wall near the ceiling."

Overall, a pretty good description of my kitchen. Now let's see if it can help me know if I should clean today. P.S. I should!

I asked, "Is the kitchen clean?" to which it replied, "Yes, the kitchen appears to be clean and well organized. The countertops are clear except for a few essential items and the dishes in the dish rack seem to be clean. There is no visible clutter or mess in the picture."



Jack and his guide dog, Baloo

I happen to know that the floors and countertops could use some cleaning. The answer is more a symptom of the fact that I took the photo of my kitchen from far away to try to capture the full room. On other occasions, *Be My AI* has reliably detected dirt in my bathtub, helped me identify entrances to buildings, read my thermostat, read ingredients on packages, and much more.

As this technology improves, I anticipate being able to share a live video feed with AI and receive real-time descriptions of the world around me. I also anticipate being able to train AI to help with navigation, avoiding bumping into things, and, of course, driving cars.

If you've not already experimented with AI, I encourage you to do so. Like the technologies that have come before it, AI is well-positioned to level the playing field for people with disabilities.

Jack McCormick graduated in 2018 from Canada's Wilfrid Laurier University in Waterloo, Ontario. He was diagnosed in high school with LCA2 (RPE65). Jack is a Hope in Focus ambassador, helping people living with LCA and IRDs. You can read his blog at jackdamccormick.wordpress.com

Events

DO YOU HAVE AN EVENT YOU WANT TO SHARE? LET US KNOW!
Email info@hopeinfocus.org with the information and a link.

NPHP5 Associated Retinal Degeneration Webinar January 2024

hopeinfocus.org

Researchers from the Foundation for the National Institutes of Health Bespoke Gene Therapy Consortium and the University of Pennsylvania discuss their *NPHP5* Associated Retinal Degeneration clinical trial in a webinar hosted by Hope in Focus. Please check our website for details.

National Institutes of Health (NIH) Rare Disease Day

February 29, 2024 • Videocast

ncats.nih.gov/rdd

More than 7,000 rare diseases affect an estimated 30 million people in the United States and more than 300 million globally. The annual NIH Rare Disease Day event features a videocast airing 9 a.m. ET, Feb. 29, 2024, to raise awareness about these disorders and the people they affect, and about the NIH collaborations advancing research for new treatments. Sponsored by the National Center for Advancing Translational Sciences (NCATS) and the NIH Clinical Center, the event features panel discussions, rare disease stories, and more.

Informa • Rare Disease Summit

March 19–21, 2024 • Philadelphia

Pre-conference workshop March 18, 2024

informaconnect.com/rare-disease-summit

Unite with the rare disease community to create powerful partnerships, advance orphan therapeutics, and deliver life-changing breakthroughs. The summit connects key stakeholders to drive therapeutic progress, propel commercial strategies, and inspire advocacy with an impact.

— SAVE THE DATE —

Foundation Fighting Blindness VISIONS Conference

June 21–22, 2024 • Chicago

www.fightingblindness.org/events/visions-2024-503

The Foundation's national conference features sessions on research, advancements, practical adapting and thriving, and opportunities to connect with the blind and low-vision community. A dynamic actionable program and agenda have been developed in collaboration with the Orphan Disease Center of the University of Pennsylvania.

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To learn more about Hope in Focus, visit www.hopeinfocus.org.

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