HOPE in FOCUS
Seeing a cure for blindness
Supporting the LCA and rare retinal disease community

SEEING HOPE | Newsletter

P.O. Box 705 | Ledyard, CT 06339 | info@hopeinfocus.org | 860-266-6062 | www.hopeinfocus.org

September 2025 | Issue 27

From the Director

Courtney Coates

Dear Friends,

What an incredible time we shared at the 2025 LCA Family Conference! It was inspiring to see individuals, families, researchers, and advocates come together with the shared purpose of supporting one another while pushing toward a future filled with more hope, answers, and progress. From powerful presentations to meaningful connections, this year's conference reminded us of the strength and resilience of the LCA community.

Looking ahead, we're excited to continue the momentum with our tenth Dinner in the Dark! This unique evening is a celebration of our community and a vital opportunity to raise awareness and support for ongoing research and resources. We can't wait to gather again for an experience that is both impactful and meaningful.

Finally, we are deeply grateful for the wave of research and treatment progress we're seeing for LCA. With multiple treatments in development, the landscape of possibilities is changing, and it's happening because of your continued involvement, advocacy, and support.

Thank you for being part of this journey. Together, we are moving closer to brighter futures for all individuals and families affected by LCA.

With gratitude,

Courtney Coates

Courtney Coates,
Executive Director

Lee St. Arnaud, Elle St. Arnaud, Jack Morris, Leslie Morris

Living with IRDs: *Insights and Inspiration from the Front Lines*

By Katherine L. Kraines, MS

The 2025 LCA Family Conference in Minneapolis, MN, offered four informational panel sessions, where the 135 attendees learned about living with Leber congenital amaurosis (LCA) and other inherited retinal diseases (IRDs), participating in clinical trials, and the latest research and therapy updates.

The first session, "Living with an IRD," was moderated by Laura Manfre, co-founder and Board Chair of Hope in Focus. This session is often a highlight for families wanting to learn how to support their children diagnosed with an IRD.

Laura posed questions to the four panel participants, which included 18-year-old Elle St. Arnaud, who has LCA *IQCB1/NPHP5* and is attending Boston University this fall, and her father Lee St. Arnaud, and 22-year-old Jack Morris, who has RP *CRB1* and is a recent graduate of Brown University, and his mother Leslie Morris.

On the next page are some of the panelists' edited responses to Laura's questions.

Continued on page 2

Living with IRDs:
Insights and Inspiration from the Front Lines

Continued from page 1

What has your school journey been like? What challenges have you faced, and how did you advocate for yourself?

Elle St. Arnaud > Elle was educated in the public schools with a paraprofessional supporting her in class from preschool through fourth grade. She said that a key to her success was learning to advocate for herself early on by participating in her individualized education plan (IEP) meetings, explaining her accommodations to teachers, and taking responsibility for communicating her needs, especially regarding her vision. Elle reflected on the importance of self-advocacy and how it has shaped her life.

"It's vital for kids to be able to explain their accommodations, as others often don't fully understand visual impairments," she said. "While I've had positive experiences, I've also faced challenges, such as being discouraged from joining the debate team or taking advanced classes. I pursued both anyway, proving that being blind doesn't limit one's ability to succeed. Self-advocacy and standing up for your needs are essential life skills."

Jack Morris > Jack started in the public schools and later switched to a private school. His vision was relatively good throughout most of his schooling. Jack thought he might have done even more to advocate for himself. Still, regular meetings with teachers and advisors proved very helpful. He said, "When people understand what you need, they generally want to help." Although at the university level, he said some professors were resistant to making accommodations.

He stressed that "Accommodations are not favors—they are necessary for success. Blind individuals are not less intelligent; they simply need equal access to materials to perform just as well as others. Having confidence in advocating for these needs is crucial, and legal protections like the ADA support this."

As parents, can you share a little about your journey? How was it seeking a diagnosis, and what are some things you wish you'd known earlier?

Lee St. Arnaud > Lee explained that Elle and her older brother, Patrick, both have LCA. When trying to diagnose Patrick's vision issues, he shared how difficult it was when the doctor offered only a brief response, instructing them to return in five months to confirm their son's blindness. Overwhelmed, the family immediately reached out to resources like the Chicago Lighthouse for the Blind, which connected them with supportive networks and foundations.

Lee stressed the importance of parents being proactive and finding tools like a detailed year-by-year visual goals guide, which they used effectively in IEP meetings to advocate for their children's needs. He emphasized the importance of being prepared, pushing for necessary services, and actively advocating for the child and family.

The St. Arnauds encouraged their children to participate in outside activities like adaptive climbing and the live hockey association. "So many times we were nervous and sweating bullets. But by the time we got done, the kids were like, you know what? We can do this!" Lee said.

Leslie Morris > Leslie said that Jack's diagnosis with a recessive form of RP at age five came as a shock because there was no family history. She described feeling heartbroken and overwhelmed by a sense of loss and isolation. Despite these emotions, Leslie and her husband, Jason, chose not to accept discouraging responses from doctors. Instead, they took action—connecting with others affected by IRDs and becoming involved with the Foundation Fighting Blindness, where both serve as trustees.

Advocacy became like a second job for the Morris family, as they worked to ensure Jack had every opportunity for a full and meaningful life. Leslie said they didn't want the disease to define their son, and they encouraged him to try various activities like baseball, football, swimming, wrestling, and guitar. "We focused on letting Jack decide when to step away from activities while gently steering him toward pursuits that offered long-term fulfillment," Leslie said.

How do you give your child enough independence to grow and to learn, while also wanting to protect and keep them safe?

Lee St. Arnaud > Lee felt that they didn't experience significant disagreements with Elle or Patrick regarding their ability to self-manage. He stressed the importance of balancing caution with encouragement and supporting early opportunities for independent experiences in the community without rushing the process. He also reflected on the importance of understanding your child's interests as they mature.

Elle St. Arnaud > Elle immediately spoke up after her father, expressing her childhood frustration and conflict with her parents over having to take Orientation and Mobility (O&M) training, particularly the inconvenience and social discomfort of being taken out of school. Despite hating it at the time, she admitted the lessons had long-term value, especially in promoting independence. She noted the

importance of starting children early with independent tasks at home, like cooking and laundry, while pacing community-based skills more carefully. "I think parents should keep their children engaged in O&M even if the child resists," Elle said. "Because it equips them with critical life skills—such as safely crossing the street—that pay off later."

Leslie Morris > Leslie emphasized the importance of parental teamwork and unity, particularly when supporting a child's journey toward independence. She admitted to being a bit of a helicopter mom. "My husband, Jason, and I agreed on things before letting Jack go out and try them," she said. "Having somebody by your side through this journey is very important." She added that when parents are aligned in decisions and approaches, it helps minimize conflict and makes the process smoother.

Jack Morris > Jack didn't recall having major conflicts and expressed gratitude for the trust his parents placed in him. One slight regret he had was not wearing sunglasses earlier, due to self-consciousness. Jack said he appreciated the opportunity to try things—even fail—while still feeling supported and safe.

Much to the amusement of the conference attendees, Jack recounted a brief period when he was legally allowed to drive, a decision made with medical guidance that, in hindsight, seemed very questionable.

"I appreciated that my parents trusted me to be able to take care of myself. This journey makes you resilient, tough, and resourceful," Jack said. "Kids can be very clever in planning solutions to their unique circumstances, and my parents trusted me in that."

Community members have expressed that mental wellness and accepting vision loss can be difficult. Elle and Jack, what are your thoughts?

Elle St. Arnaud > "Not wanting someone to see your pain and not wanting to wear sunglasses or use a cane will likely peak around middle school," Elle said. "No one wants to be different, and you want to be 'on trend' with everybody else."

She added that having a para in the classroom with her all of the time was an inhibitor and made it harder for her to talk with other kids. "Parents should try not to hover when their child is with other children because it can make them feel less confident or social," she said.

Elle said she made it a priority to share her experiences as a blind person with her sighted friends. "We'd make jokes about it, and I talk about it constantly. I'd share what I've experienced and every blind story I've had," she explained. "I think that's important because when you need to ask your friends for help, they are a lot more understanding because you've explained it and talked about it so many times with them."

For example, she told her friends about walking into a door, which helped them be more aware that she might not see it, allowing them to take the initiative to guide her when needed. "I think the easiest way to be accepted is to talk more about your blindness so it feels more normal," Elle said.

Jack Morris > "I think there's a line to be found between letting a disease define you and rejecting that it's a part of you, because retinitis pigmentosa (RP) has been a huge part of my story—a beautiful but also a tough part. It's something that feels different and lonely, but also unifying and community-building," Jack said. "If we can find ways to live in and with our disease rather than despite it, that's where truth and freedom lie for me."

He offered advice to parents, saying that they needed to be strong in ways that aren't obvious. "One of the hardest things for parents is watching their child struggle. But sometimes the biggest strength is allowing them to struggle. Kids need to feel the full spectrum of emotions—challenge, loneliness, empowerment, and togetherness," Jack said. "If parents can be there as a foundation, even when things are hard, and let them be hard before stepping in with action, that's one of the most powerful and meaningful things they can do for their children."

If you, or someone you know, has an inherited retinal disease due to mutations in the *IQCB1/NPHP5* or *CRB1* gene, email info@hopeinfocus.org to join our contact database so we can give you up-to-date happenings with your gene.

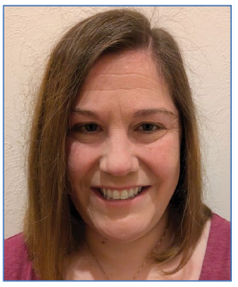
MOVING FORWARD:
Understanding More about Clinical Trials

By Katherine L. Kraines, MS

The second panel session of the 2025 LCA Family Conference, "Participating in a Clinical Trial," examined clinical trial participation from the researcher and patient perspectives. This session supports a Hope in Focus goal to educate the Leber congenital amaurosis (LCA) community so members are ready to participate in clinical trials. For researchers, informed and prepared LCA patient groups are critical to moving a new therapy through the testing pipeline.


Ben Shaberman


Tomas Aleman, MD


Sarah McCabe

Ben Shaberman, vice president of Science Communications at the Foundation Fighting Blindness, was the moderator. The panelists were Tomas Aleman, MD, a researcher with over 30 years of experience in researching genetic therapies related to inherited retinal diseases (IRDs), and Sarah McCabe, a mother and teacher, who has the *RPE65* gene mutation. Sarah participated in a gene therapy study in 2007, and 14 years later was treated with LUXTURNA®.

Drug Development

Ben provided an overview of the drug development process, highlighting its complexities and length—often taking 10–15 years—and costing tens of millions of dollars. For retinal diseases like LCA, the development of a drug or therapy begins with identifying and understanding the mutated gene causing the degeneration.

Transitioning from animal or lab models to human trials is a significant hurdle, requiring higher-quality manufacturing standards, regulatory compliance (e.g., FDA), considerable funding, and specialized expertise. "This phase, called translational research, is often referred to as the 'valley of death,'" Ben said. "Because many therapies fail to progress to clinical trials."

LCA Gene Research

Dr. Aleman co-directs the Center for Hereditary Retinal Degenerations (CHRD) at the Scheie Eye Institute at the Perelman School of Medicine, University of Pennsylvania, where his groundbreaking work has transformed the treatment of LCA. "Unlike what many people believe, most LCA patients are not completely blind, and their retinas often remain structurally intact," Dr. Aleman said. "This makes the condition an ideal candidate for experimental therapies."

Early research focused on *RPE65*-related LCA and started with animal models that helped pave the way for clinical applications. After a decade of preclinical research, Dr. Aleman's team moved into human trials, with LCA patients like Sarah McCabe playing a vital role.

Clinical trials require long-term patient commitment and rely heavily on funding from smaller biotech companies. Patient selection for trials is strategic—those chosen typically have structurally preserved retinas with poor function, maximizing the likelihood of measurable improvement. Dr. Aleman emphasized that exclusion from a trial does not mean the therapy won't eventually be available for an individual; rather, it reflects the strict criteria needed to answer key safety and efficacy questions. Looking ahead, Dr. Aleman's goal is to expand the proportion of treatable LCA forms from roughly 25 to 50 percent.

Doing Gene Therapy

Dr. Aleman explained that the gene therapy process for treating IRDs focuses on subretinal delivery, with the surgery resembling retinal detachment repair. The procedure is performed under general anesthesia and only takes 5–10 minutes. It is done by entering the eye through three small incisions, removing the gel-like vitreous, and using a hair-thin needle to deliver the gene therapy.

Following surgery, a rigorous monitoring process assesses the treatment's safety and effectiveness. This process includes frequent follow-up visits in the early stages—and then at one, three, and six months and a year—during which visual function is tested and retinal imaging is conducted.

Conference attendees listening to the "Participating in a Clinical Trial" panel session.

Participation in gene therapy trials is entirely voluntary, and patients can choose to withdraw at any time. However, once the gene therapy is delivered, it cannot be undone. The therapeutic genes remain in the eye's cells indefinitely, making informed consent and long-term commitment critical components of the clinical trial process.

"While gene therapy offers transformative potential, it also requires thoughtful implementation, long-term follow-up, and a commitment to tailoring support beyond the surgical intervention," said Dr. Aleman. "The mission is not only to restore vision but to improve the quality of life and long-term outcomes for patients, especially children, by intervening as early as possible."

Sarah's Story & Clinical Trial Experience

When Sarah was about ten days old, her mother, an ICU nurse, noticed that she wasn't following the developmental patterns she'd observed with her first child. Concerned, they visited a pediatrician who suspected something was wrong with Sarah's vision. Further evaluation by a neurologist ruled out any neurological issues, and her parents were assured that Sarah would hit her developmental goals.

Regular eye exams ensued, with her parents keeping detailed records. Eventually, Sarah was referred to the University of Iowa, where tests suggested LCA, but at that time, genetic testing wasn't available. A definitive LCA genetic diagnosis (*RPE65*) was finally made when she was in eighth grade.

At age 19, she was recruited for a clinical trial after struggling with vision during college. After going through the initial interviews and assessments, she was approved as a clinical trial participant. Sarah said the doctors clearly explained all the details and risks of the trial and that it was an experimental procedure primarily aimed at testing safety. The decision whether to move forward was left up to Sarah and her family. "There wasn't a lot of talking with my parents about it," she said. "We knew things weren't going to get any better if I didn't participate, and I could be a part of helping [research advance].

Toward the end of her senior year, her family drove her from Iowa to the University of Florida, where she underwent her first gene therapy surgery, as part of the clinical trial, at age 23, describing it as terrifying but perfect.

During the surgery, Sarah was awake—a protocol that has since changed, with patients now put under general anesthesia. Post-surgery, she was able to read a giant letter "A" on a card, confirming that the surgery hadn't worsened her vision. Over time, she noticed a new visible area in her field of vision, referred to as a "headlight," which was a significant improvement.

Sarah's recovery involved staying in Florida for a month with her family, with frequent follow-up visits stretching out from monthly to annually. Her clinical trial team remained in contact with her years after the trial formally ended.

Fourteen years later, after LUXTURNA® was FDA-approved, she received the gene therapy at the University of Iowa, which improved her vision. Now in her 40s, Sarah's primary goal is to maintain the stability of her vision. She summarized her clinical trial and gene therapy experiences, saying, "It was a long time ago now, but it was a very cool experience. All of it!"

Reports on Gene Therapy Advances: *A Highlight from the 2025 Hope in Focus Conference in Minneapolis*

Ben Shaberman
VP, Science Communications, Foundation Fighting Blindness
Advisor, Hope in Focus

Gene therapy is unequivocally the most advanced approach for treating retinal diseases like Leber congenital amaurosis (LCA). Of course, there's LUXTURNA® which is FDA-approved and has restored significant vision for people with LCA caused by *RPE65* mutations. But several other emerging gene therapies are, or will soon be, in clinical trials. Excitingly, some are restoring vision early in human studies.

I had the honor and privilege of moderating an expert research panel at the 2025 Hope in Focus Conference last June in Minneapolis to discuss some of the exciting developments in LCA gene therapies. The three panelists were Kenji Fujita, MD, chief medical officer, at Atsena Therapeutics; Sarah Tuller, JD, chief regulatory officer at Opus Genetics; and Bikash Pattnaik, PhD, a professor at University of Wisconsin-Madison.

Atsena's LCA1 Gene Therapy Moving into Phase 3

Dr. Fujita delivered the exciting news that Atsena's LCA1 (*GUCY2D*) gene therapy performed very impressively in a Phase 1/2 clinical trial. "We were super-thrilled with the results," he said. "The gene therapy worked better than we expected." Thanks to the excellent results, the gene therapy is moving into Phase 3 in a co-development partnership with Nippon Shinyaku which brought a few of their representatives to Minneapolis.

The Phase 1/2 trial enrolled nine adults in Part A (the dose escalation group) to evaluate initial safety and determine the optimal dose. An additional three adults and three pediatric patients were subsequently dosed. Patients receiving the highest dose (all were treated in one eye) had 100-fold improvement in retinal sensitivity, as measured by full-field sensitivity (FST). Some had 10,000-fold improvement. Patients were also able to navigate a multi-luminance mobility test (MLMT) in dimmer light (two lux levels lower) after treatment. "This was a transformative difference, on par with what we have seen with LUXTURNA," said Dr. Fujita.

The Phase 3 clinical trial will enroll a larger group of patients and treat both eyes. Some patients will be in a deferred treatment group, serving initially as controls.

The Foundation Fighting Blindness, through its RD Fund, is an original investor in Atsena.

Opus Reports Vision Improvements in LCA5 Gene Therapy Clinical Trial

Opus Genetics, a company established by the Foundation Fighting Blindness in 2021, launched its first clinical trial in 2023 for an LCA5 gene therapy. LCA5 is a severe retinal degeneration diagnosed in a child's first year. It is also very rare, affecting only about 200 patients in the US.

Opus reported excellent results for the first three patients (adults) in the trial with improvements in FST and virtual maze navigation. The company is now dosing pediatric patients and expects to report on them in the third quarter of 2025. "We are trying to move forward as aggressively as the FDA will allow," said Ms. Tuller.

She acknowledged the great work of Dr. Tomas Aleman, the principal investigator on the trial, who was also at the meeting and had an engaging discussion with Sarah McCabe, one of the first patients to receive an *RPE65* gene therapy.

A CRISPR Therapy is Emerging for LCA 16

Dr. Pattnaik reviewed his team's emerging CRISPR gene editing approach for correcting the W53X mutation in the gene *KCNJ13* which causes LCA16. He explained that the treatment works like molecular scissors to cut out the mutation.

Dr. Pattnaik is using lipid nanoparticles—which are like microbubbles—to deliver the treatment into retinal pigment epithelial (RPE) cells. Unlike most other genetic therapies which use engineered viruses to get genetic cargo into cells, nanoparticles have the advantage of being able to deliver therapeutic cargo of any size. Also, they are less likely to cause an immune reaction than viral systems.

Dr. Pattnaik tested the approach in cells and small animal models, and is now evaluating it in a large animal. He said the FDA is very positive about their current development plan.

The CRISPR therapy is currently funded through a grant from the National Institutes of Health (NIH) and was previously supported by the Foundation Fighting Blindness.

Dr. Pattnaik is also a co-founder of Hubble Therapeutics which is advancing a *KCNJ13* gene augmentation therapy developed in his lab.

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

GUIDING LIGHTS: *The Transformative Role of Mentors*

Jack and his guide dog, Baloo

When I was 16, my impaired vision started to get worse. I was graduating from high school soon and didn't know what these changes would mean for my future. I had dreams of becoming a CEO. As my vision worsened, I was more aware of the accessibility and attitudinal barriers I would need to overcome—not to mention adjusting to declining vision.

It was a challenging time. I was worried that the barriers were too significant to surmount. At times, I questioned if it was even worth trying. In the darker moments, I told myself to give up. Thankfully, there was a part of me that wasn't willing to do that, and I realized that I wasn't the first blind/visually impaired guy with big dreams. Maybe I could find someone who had done it before?

Dennis, Jack, and Baloo

Then, I found Dennis Atkins' website. He was exactly who I needed. Dennis is a CEO with a PhD who raises cattle and runs marathons. He is a man who gives so much to his community and who supports his family. Did I mention he is blind?

I was a scared teenager, and I sent him what I can only describe as a desperate email. He replied with kindness and optimism, offering to do what he could to help me. Dennis provided advice and encouraged me to continue with my post-secondary plans despite the hopelessness I felt.

I shared my fears and desire to change the barriers that people like me experienced. He told me not to forget my other dreams while I volunteered with disability organizations in an effort to change some of those barriers. His mentorship helped me through one of the darkest periods of my life.

I continued to receive support from Dennis, reaching out to him when I thought he could offer assistance. While in college, I co-founded a student club that promoted disability inclusion on campus, and Dennis flew to Canada to speak at one of our events. He is one of the most incredible people I've had the pleasure of knowing.

Years later, Dennis and I still keep in touch. But he has not been my only mentor. I've had many mentors for the different seasons of my life. When I feel lost, stuck, or want to grow, I look for mentors who can share their wisdom, helping me take on the next challenge.

I don't know where I'd be without Dennis. Would I have given up without someone to look up to? At the very least, my life would have been harder. I know I would not be the person I am today without the mentors I've had in my life. Living with a visual impairment isn't easy. It's important to know that you are not alone and that there are many people like Dennis. When you encounter a barrier or want to take on a new challenge, take the initiative to find people you can reach out to for support. You will find someone who can help, even if it is just a bit. Life is better and richer when you've got a mentor cheering you on.

Our Family Connections program makes it easy for those with LCA or another rare inherited retinal disease to connect with another family or individual by phone or email. Details: ***hopeinfocus.org/for-families/connect***

Dennis Atkins' Website: ***dennisatkins.com***

Jack McCormick is a human resources professional working in the tech sector. 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 more about him on his LinkedIn profile by scanning the QR code to the right.

Events

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

Dinner in the Dark

November 1, 2025 • Foxwoods Resort Casino, Mashantucket, CT

hopeinfocus.org/dinner

Dinner in the Dark, our primary fundraiser for the year, 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. Get ready for an incredible evening that is a lively sensory adventure with a stellar menu, fine wines, and more!

VisionWalk — Foundation Fighting Blindness

www.fightingblindness.org/visionwalk

Since its inception in the Spring of 2006, VisionWalk has raised over $71 million to fund sight-saving research. Join a VisionWalk in your community! Together, we step closer to fighting blinding diseases.

NORD Breakthrough Summit

October 19–21, 2025 • Washington, DC

nordsummit.org

Shape the future of rare disease treatments, research, and policy at the NORD® Rare Diseases and Orphan Products Breakthrough Summit.® Together, we can advance innovation for the more than 30 million Americans—and more than 400 million people worldwide—with rare diseases!

HOPE in FOCUS

P.O. Box 705 | Ledyard, CT 06339

The *Seeing Hope* Newsletter
is published quarterly by Hope in Focus, a 501(c)3 patient advocacy organization dedicated to generating awareness, raising funds for research, and providing education and outreach to the LCA and rare inherited retinal disease community.

Learn more at hopeinfocus.org

***Seeing Hope* Newsletter Team**
- Courtney Coates, Executive Director
- Katherine L. Kraines, MS, Communications Manager
- Genevieve Orcutt, Marketing and Community Engagement Manager
- Gina Morin, Graphic Designer

This newsletter is made possible by the generosity of:
- Opus Genetics
- MeiraGTx
- Atsena Therapeutics
- Sepul Bio

The wood fiber used to make this paper is independently certified to come from responsibly managed forests.

NON PROFIT ORG
US POSTAGE PAID
MYSTIC, CT
PERMIT NO. 16