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

SEEING HOPE | Newsletter

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MY FRIEND LUCA: *Balancing Hope and Reality*

By Katherine L. Kraines, MS

Blair, Luca, and John Corso

Three-year-old Luca Corso's preschool teacher announced that everyone should quickly sit on their floor shapes for carpet time. As the other children eagerly scattered to their assigned spots, Luca wandered. He wasn't being disruptive—he just wasn't sure what to do. Even with glasses and a cane, he couldn't locate a flat shape on the floor. Luca's vigilant parents, Blair and John Corso, along with his early intervention team, got involved, and the preschool quickly provided him with a small chair for carpet time.

Diagnosis Shock

Luca was diagnosed with the recessive *AIPL1* gene mutation that causes Leber congenital amaurosis 4 (LCA4) when he was about 14 months old. According to the Foundation Fighting Blindness, this rare but severe form of LCA affects "only a few hundred people in the US and less than 10,000 people globally." The news was devastating to first-time parents, Blair and John, and almost unbearable when they were told that their little boy would likely lose all of his vision by age four.

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FROM THE DIRECTOR

As 2025 comes to a close, we are filled with gratitude for our supporters, the resilient and inspiring Leber congenital amaurosis (LCA) community, and the many accomplishments achieved together this year. What a year it has been!

We just hosted another successful Dinner in the Dark, which brings meaningful awareness and critical support for our mission. This event is a powerful reminder of the challenges faced by individuals living with vision loss and the importance of coming together to illuminate a path toward effective treatments.

June brought our LCA Family Conference, where families, researchers, clinicians, and industry partners gathered for two unforgettable days of learning, connection, and hope.

This year's momentum reflects the strength of our mission and the trust you place in us. With a growing staff and an expanding network of partners and community members, we are more energized than ever.

As we look ahead to 2026, we are excited to grow our programs, deepen our outreach, and expand the ways we educate, empower, and connect individuals and families living with LCA and rare inherited retinal diseases. With your partnership, we will continue advancing toward treatments while supporting our community every step of the way.

Thank you for your steadfast support. We cannot do this without you.

Courtney Coates

Courtney Coates,
Executive Director

MY FRIEND LUCA:
Balancing Hope and Reality

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A Ray of Hope

The family was given a ray of hope after hearing about a new LCA4 treatment developed by MeiraGTx that showed efficacy in a trial involving 11 children in the United Kingdom (UK). The therapy, which uses a human-engineered adeno-associated virus (AAV) to deliver copies of the therapeutic gene into remaining photoreceptors, has not yet been approved for use in the UK or the United States. If successful, the treatment could restore some of Luca's vision.

Hoping beyond hope that the treatment becomes available before Luca turns four, the Corsos know that his window of opportunity is rapidly closing. Another potential hurdle is the cost. "Not only does the treatment require approval, but we don't know the cost, since the trial would no longer fund it," Blair said.

Balancing Emotions

For Blair, the news of the UK trial's success was at once wonderful and disheartening. "I saw one of the boys who had the treatment and spoke with his mother," she said, choking back tears. "After the treatment, he could see facial expressions. It would mean everything if Luca could experience that too. When I read stories about the children treated, it struck me that being able to see things like that could really help Luca interact with us and his peers."

The Diagnostic Journey

When Luca was born, the Corsos were living with Blair's parents while they searched for a house. It was then that they first suspected something was wrong with Luca's vision. "My Dad kept wondering why Luca's eyes were going back and forth so much," Blair said. "Was it normal?"

The pediatrician was also concerned about their four-month-old son's eye movements (nystagmus) and quickly referred Luca to a local eye doctor. After confirming Luca's vision loss but seeming less worried about the nystagmus, the eye doctor referred the family to Mays Al El-Dairi, MD, a pediatric ophthalmologist and neuro-ophthalmologist at Duke University Eye Center.

"Dr. El-Dairi examined Luca and mentioned the possibility of LCA and recommended genetic testing with Ramiro Maldonado, MD, a retinal specialist at Duke," Blair said. Dr. Maldonado arranged the genetic testing and wanted to do an electroretinography (ERG), a diagnostic test that measures the electrical activity of the retina. But Luca needed to be at least a year old to have the ERG.

Test Results

"Luca was about 14 months old when the ERG was done, and it showed that he had about 50 percent of his vision. The genetic test indicated LCA4, and Dr. Maldonado said that our toddler would likely be blind by age four," said Blair. "It was a very rough day. John was at home sick, our dog had just died, and my mother and I were in the surgery waiting area, bawling our eyes out." Luca was Dr. Maldonado's first case of LCA4.

Luca walking with his Grandfather

Help!

Like all parents hearing an LCA diagnosis, the Corso family was desperate for more information and support. They immediately began looking on Facebook, where they discovered Hope in Focus (HIF). Through HIF, they started connecting with the LCA community, and Blair was paired with an HIF Ambassador, Ashlyn, whose young son has LCA10.

"I can pick up the phone or shoot a Facebook message to her, and I don't have to explain things," Blair said. "It's wonderful to have the support, especially when you don't know what you are doing. If I have a question, Ashlyn offers input or helps me reach out to someone else. It's like a big family of support, and it's so encouraging to know that other children with LCA are thriving."

Early Intervention

Early in Luca's diagnostic journey, the local eye doctor suggested pursuing 'early intervention' due to his vision loss. Unsure what 'early intervention' meant, the Corsos learned more about it and reached out for help, a decision that would prove crucial in Luca's journey.

The Corso's live in North Carolina, where early intervention is available through the state's Children's Developmental Services Agencies (CDSAs). "They sent some people out to talk with us about LCA,

Blair and Luca

John and Luca

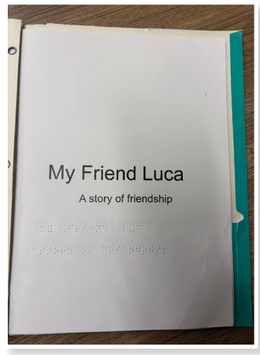
Luca on his mat

and there was an actual team involved. We had a caseworker, a vision teacher, and an orientation and mobility coach," Blair said. "They were fantastic in guiding us about how to teach Luca and helping him keep on track with his learning."

Blair stressed the importance of accessing early intervention and said, "Our CDSA team visited us before Luca was in preschool and worked with him at least once a week. When he began preschool, the team went there as well, helping the teacher to understand his needs, such as where to position him so he could see in the classroom."

The team also conducted many in-home learning sessions. "They suggested different things to support Luca in school and at home. His occupational therapist recommended that we help him learn how to move to songs so he wouldn't stand motionless while his classmates danced," Blair said. "You know, it's not always obvious or intuitive for parents to know what to do!"

One of his vision teachers, Ms. Charli, created a book for his preschool called "My Friend Luca," which explains that Luca's eyes don't work well and that he uses his hands to see the world. "It helps other children understand why Luca might want to hold their hand or reach out and touch their face," Blair said.



Ms. Charli's book created for Luca's classmates

There were also monthly trips into the community with his orientation and mobility coach, Ms. Annette. "We went to the strawberry patch, the grocery store, and Lowe's," Blair said. "We did normal things so she could observe him in public—such as how he navigated with his cane."

Luca's current mobility and orientation coach, Mr. Mike, is the past president of the Maryland School for the Blind and a member of the local school system that now serves Luca's needs. "Mr. Mike is just amazing!" said Blair. "He focuses on learning through play and wants Luca to think of him as a fun grandpa. He even got Luca to sit on the swing, which he was terrified to do."

Going Forward

The Corso's attended this year's Hope in Focus Family Conference for the first time and found the information and variety of speakers very helpful. Meeting other families with children who have LCA was supportive, especially those dealing with the same gene mutation and its consequences.

Blair and John recommend that parents seek early intervention resources for their child with LCA and be open to help. They hold out hope that the treatment in the UK might become available for Luca while continuing to prepare him for a future where he is blind.

For now, the couple finds great joy in their little boy and his "can-do" spirit, delightful personality, and bright, engaging mind. They have and will continue to ardently advocate for Luca as he walks into the future surrounded by their unending love and support.

For information on the treatment for LCA4, scan this code.

If you, or someone you know, has an inherited retinal disease due to mutations in the *AIPL1* gene, email info@hopeinfocus.org to join our contact database so we can give you up-to-date happenings with your gene.

Thank You

Your support at

DINNER IN THE DARK

helps us continue serving and supporting the LCA community.

40 SPONSORS

RAISED OVER $190,000

260 ATTENDEES

25 AUCTION ITEMS

and COUNTLESS photos with the zebra!

Photos by PubliCreatives

Promising RNA Therapies for LCA10 and USH2A Move Back into Clinical Trials

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

FOUNDATION FIGHTING BLINDNESS

Moving emerging therapies through clinical trials and across the finish line is often challenging—and in some cases, harrowing. Many treatments never make it.

In early 2022, the future looked bleak for ProQR Therapeutics' two RNA therapies in clinical trials. The biotech company reported that sepofarsen, its RNA therapy for LCA10 (IVS26 mutation in *CEP290*), did not meet its primary endpoint of improvement of at least three lines in best-corrected visual acuity or BCVA. (Improvement in BCVA was only on average two lines in the Phase 2/3 trial.) That news came despite vision improvements, some significant, for many patients. The endpoint misled ProQR to stop development of its ophthalmology assets—sepofarsen and ultevursen (exon 13 mutations in *USH2A*)—and attempt to find a company to acquire them.

Mike Schwartz, who was then vice president, global project leader, at ProQR, said, "That was devastating for me, the doctors, and the patients." He noted that one patient with LCA10 in the trial with only light perception gained enough vision after receiving sepofarsen to see letters on an eye chart. Another LCA10 patient in the study returned to his work as a carpenter after treatment.

Fortunately, a year and a half later, the large European eye care company Théa acquired sepofarsen and ultevursen and formed the Sepul Bio business unit to move the therapies back into clinical trials. Many former ProQR staff went to Sepul Bio, including Mr. Schwartz, who is now their chief operating officer.

The global HYPERION Phase 3 clinical trial for sepofarsen and the LUNA Phase 2 trial for ultevursen are now underway. Using what was learned from the ProQR trials, the Sepul Bio team made significant changes to the designs (protocols) for the clinical trials, changes they believe will greatly improve chances for success. Mr. Schwartz thanked the Hope in Focus team for providing input from patients for the sepofarsen clinical development program.

One major change in the new sepofarsen clinical trial protocol has to do with the placebo. In most clinical trials with regulatory authorization, the treatment group is compared to a placebo or control group to ensure that efficacy is indeed a result of the treatment. In the original sepofarsen trial, treated eyes of LCA10 patients were compared to the eyes of untreated LCA10 patients (i.e., the control group). Comparing treated patients to untreated patients was less than ideal because of significant variations in vision loss among LCA10 patients. So, in the new trial, each LCA10 patient will have one eye injected with sepofarsen and the other will get a saline placebo injection. The patient won't know which eye is getting the treatment. Sepul Bio believes comparing untreated and treated eyes for the same patient will lead to less variation and a stronger efficacy signal.

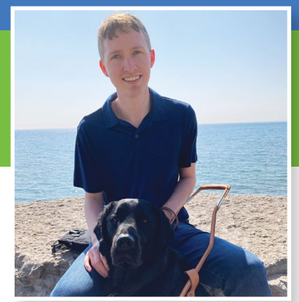
Keep in mind that sepofarsen injections are made into the vitreous, the soft gel in the middle of the eye. These intravitreal injections are performed routinely (e.g., monthly) and safely in doctors' offices for treating age-related macular degeneration. In the sepofarsen clinical trial, patients will receive injections every six months.

Sepul Bio's RNA therapies, known as antisense oligonucleotides (ASOs), are tiny pieces of genetic material that fix mutations in RNA—the genetic messages that cells read to make proteins critical to the cells' health and function. Stay tuned. We will report on updates from the trials as soon as we receive them.

For more information on the sepofarsen or ultevursen trials, send an email to: **contact@sepulbio.com**.

Jack McCormick column

TESTING THE LIMITS: *What is your Kilimanjaro?*



Jack and his guide dog, Baloo

Throughout my life, I've pushed my body to do some incredible things—from wrestling in high school to running a marathon to later backpacking in the wilds of British Columbia. But on October 7, 2025, I literally reached new heights as part of a group of nine legally blind individuals who successfully summited Tanzania's Mount Kilimanjaro. Our group set a world record for the largest number of legally blind climbers to reach the mountain's summit in a 24-hour period!

As the biggest mountain in Africa and the highest freestanding peak in the world, Kilimanjaro is challenging for everyone. It took seven days of grit, fatigue, and trust in our guides to reach the point where we began the final climb to the top. The physical strain left my body aching, and altitude sickness gave me one of the worst headaches I've ever had. Sleeping in a tent in freezing conditions was wearing, and I quickly missed hot showers. Within twenty minutes of starting our ascent, I lost count of the rocks I had to climb over. Being legally blind required my full concentration. I had to carefully probe the ground ahead with my hiking poles before each step and listen intently to my guide's directions, alerting me to constant obstacles along the trail.

I also experienced many challenges. On day three, I had severe dehydration and nearly fainted trying to make it to the washroom in the night. Reaching the summit meant experiencing oxygen levels as low as 47 percent, leaving me breathless every few steps. The descent was no less demanding, for hours I slid down loose gravel, followed by two days of climbing down rocky terrain on tired legs.

Getting to the top of this famous mountain was worth it, but I'd be lying if I said I wanted to do it again. You might be thinking this sounds like a great adventure and wonder how you might do something like this. Or, maybe you're wondering how we safely accomplished the climb.

The answer is simple—with a lot of support from local experts. We partnered with Nana Safaris, a tour company experienced in guiding legally blind climbers, which matched each member of our group with a local guide. My guide, Thomas, was certified by the Tanzanian government and had summited Kilimanjaro 370 times before our climb. Before setting out, we discussed how he could best assist me. Throughout the trek, Thomas carefully led the way, holding one of my hiking poles so I could sense the path ahead. He gave clear verbal directions and, in high-risk sections, had me hold on to a rock or showed me exactly where to step by placing my pole. Thanks to his expert guidance, I always felt safe.

Challenging adventures like this are possible with proper planning and support. If you're considering climbing Kilimanjaro or visiting Tanzania, Nana Safaris is the most experienced company for supporting people with disabilities. We documented our journey and will be releasing a documentary about our adventure—follow us on Facebook to stay updated.

Nana Safaris

Kilimanjaro Blind on Facebook

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. Learn more about him on his LinkedIn profile by scanning the QR code.

Events

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

Dinner at Cedars

February 27, 2026 • Cedars Steak & Oysters, Mashantucket, CT
fundraise.givesmart.com/f/5s7c/n?vid=1myxmm
Bring a friend and plan to meet new ones! Be part of an intimate dinner with 20 supporters of Hope in Focus on Friday, February 27, 2026, at 7:00 p.m. at Foxwoods Resort Casino. Enjoy a refined, upscale evening designed to feel both personal and elegant.

VISIONS 2026: United in Vision

www.fightingblindness.org/visions-2026
The United in Vision 2026 conference unites two prestigious events—the Foundation Fighting Blindness VISIONS conference and the Retina International World Congress—into one extraordinary global gathering. Scheduled from **Wednesday, June 10 to Saturday, June 13, 2026**, at the Fort Worth Convention Center in Fort Worth, Texas, this premier event brings together individuals and families impacted by blinding retinal conditions, alongside leading researchers, clinicians, patient advocates, and industry partners, to foster collaboration, innovation, and hope.

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The *Seeing Hope* Newsletter
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Learn more at hopeinfocus.org

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