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

March 2025 | Issue 25

From the Founder:

Laura Manfre

Dear Friends,

I hope your new year is off to a great start! Ours began with three new board members, a strategic plan to support our LCA community, an upcoming rare disease walk in Connecticut, and planning for the June LCA Family Conference in Minneapolis.

As I write, in early February, changes in the federal government may have immediate and longer-term impacts on LCA research and clinical trials. With budget freezes, cuts, and agency upheavals at the FDA and NIH, we're concerned about the advancement of congressionally-funded research initiatives and possible impacts on rare disease research and policy moving forward.

At Hope in Focus, we're actively monitoring these developments and collaborating with partners to navigate this evolving landscape.

See you in Minneapolis!

Keeping hope in focus,

Laura

A NEW CHAPTER: Reflections on Seven Years of Hope and Inspiration

By Angelica Bretón Morán and edited by Katherine L. Kraines, MS

Angélica Bretón Morán (right) with Dr. Emily Place (left) during the 2023 Hope in Focus Family Conference in Indianapolis

Angelica Bretón Morán, a 29-year-old musician and educator from Mexico, was born with Leber congenital amaurosis (LCA) due to *RPGRIP1* gene mutations (LCA6). In 2018, she shared her inspiring story with Hope in Focus (HIF), and since then, she has attended several LCA family conferences. We checked in with her seven years later to see how her journey has evolved. Through a Q&A, Angélica reflects on her experiences and insights.

What changes have you experienced over the past seven years?

I graduated from college in 2020 with a degree in music as a pianist, just as COVID-19 started. The plan was to take two exams—one as a soloist and another with the chamber orchestra but the pandemic delayed graduation. Graduating students were instructed to upload their performances to YouTube, and our professors evaluated them. The results were announced during an online graduation ceremony.

In October 2020, I began an online master's degree in education at Universidad Tecmilenio. At the same time, I was working on my project titled "Area of Musical Research for the Blind and Visually Impaired," which I had been volunteering on since I was age 10. It resulted in my job as the creator, founder, and leader of that department, later renamed the Music Research Center for Visual Disabilities (CIMUDIV). I completed my master's degree in 2022, became certified as a professional instructor to conduct workshops, took a leadership course, and completed a diploma in music psychology.

Continued on page 2

A NEW CHAPTER: Reflections on Seven Years of Hope and Inspiration

Continued from page 1

At ISU University, I began a doctorate in the Development and Innovation of Educational Institutions in November 2024 to create academic content about the inclusion of people with visual disabilities in professional music. This area has very little content developed by someone who has lived this reality firsthand.

Can you share more about your job?
I work at the Faculty of Music of the Universidad Autónoma de Nuevo León (UANL). As the coordinator, founder, and creator of the Music Research Center for Visual Disabilities (CIMUDIV), we make the curriculum accessible to students with visual disabilities, including creating braille sheet music and providing personalized student support. It is not a special program for blind students. Instead, students use the same materials and perform like their sighted peers in their classes. I teach musicography to students with visual disabilities so they can read sheet music, and I support their teachers when needed.

We are constantly searching for tools and strategies that make our work and the students' learning easier, which involves researching, experimenting, and adapting. It's not unusual to find people with visual disabilities who are active music community members, but much still needs to be done! Changing our mindset and that of society requires patience.

What has been encouraging?
Something that thrilled me was the Blind Concert performance held in November 2023. Because it was canceled during the pandemic, I thought we might never experience it again. The concert, held for 11 years in various locations, raises awareness through music by taking the audience on an emotional journey. For me, music is a powerful language that reaches the deepest fibers of the soul. Seeing the entire cast reunited was an unforgettable and emotional experience. In May 2024, the concert was performed outdoors for the first time before 300 people, achieving another milestone.

What are your observations related to LCA?
Learning never stops, and it is fantastic to see how the topic of LCA has progressed. We're talking about clinical trials, experimental treatments, research, and new diagnoses, things that seemed unthinkable 20 years ago. When I was younger, we weren't sure if I had LCA. Now, hearing that someone has a genetic diagnosis is incredible.

It's become easier to access the LCA community, which is more informed and has more people willing to share their experiences. The support network is stronger, and patients of all ages are involved. Social media and Facebook groups allow us to stay in touch with other families and organizations like Hope in Focus.

One exciting breakthrough is that Odylia Therapeutics is developing a gene therapy for my mutation, the *RPGRIP1* gene, and testing it on mice. As a 29-year-old blind person, the idea that my vision might improve in the future is exciting and terrifying!

What are your thoughts about the LCA Family Conferences you've attended?
My experiences at the conferences

Angélica Bretón Morán (right) with Toprak Kulekci (center) at the 2023 Hope in Focus Family Conference in Indianapolis

have been wonderful! I met many people whom I now care deeply for. At the 2023 LCA Conference onference, we met Toprak and her family from Turkey. Toprak's parents saw my story on the HIF website, and it was touching to hear that my story gave them hope. It's important for adults with LCA to attend the conference and share their experiences with other families.

We hear from the researchers, foundations, and people who make LCA research possible during the conference. At the first LCA conference, I met Dr. Emily Place, who gave me my genetic result when I was 20. I couldn't believe she was standing before me, and I hugged her with gratitude.

Meeting Toprak reminded me of another family at my first LCA conference. We met a 2-year-old girl named Dafne and her parents. Our parents started talking, and the girl and I had similar characteristics. My parents said that seeing Dafne was like seeing me when I was little, and the anxiety expressed by her parents was similar to theirs. I understood how important it was to be at the conference and how difficult it is for other families to process what I live with daily. I felt a moral commitment to be more involved in the LCA community as an adult.

At another conference, I met a mother with LCA traveling alone with her baby, who did not have LCA, leaving her other children at home. I was a teenager and had never met a mother with LCA. This woman became a role model for me.

I've learned the most about LCA at the conferences. There, we can ask our questions directly to professionals, listen to others' concerns, and learn from their stories. I've also seen how the LCA community has grown in numbers and knowledge. Moreover, there's yet to be published scientific information shared, which feels like privileged access.

Attending the LCA conference feels like reuniting with a big family. I am deeply grateful to HIF for all the support they've given. Every second I've spent at the conferences has been worth it.

How do you bring conference content back to your LCA community?
Whenever someone contacts me, I tell them about HIF and ask them to follow its social media. I also share my conference experiences and explain why attending and getting involved in the community is so important.

Our Spanish-speaking WhatsApp group has people from different parts of the world. We share what is discussed when I attend conferences or when a group member attends other LCA-related events. Many members speak only Spanish, so I support them when they need translations, but I also encourage them to write in Spanish in the Facebook groups. Many people from other countries post in their languages, but we can translate texts easily thanks to technology.

Also, artificial intelligence and technology are quickly advancing, so the language barrier is now less complicated. I encourage people not to let language be a limitation. My dad and I started attending these conferences when we spoke little English and didn't understand much. That experience pushed us to improve our skills.

You are an ambassador for LCA in Mexico. How did that evolve, and what did it look like?
When I was young, no community or information about LCA existed in Mexico. My parents were very worried about my development and diagnosis. However, one day, they read a letter from a 22-year-old Italian girl with LCA who studied music, giving them great hope.

When I was 22, I wrote a post on my Facebook blog about my experience with LCA and mentioned not knowing other people with my genetic mutation. The response was unexpected! Many people and organizations shared my story, and that's how I met people from Mexico and Spain.

Most of the people with LCA who reach out to me do so via social media or through the families in our WhatsApp group. Using WhatsApp, we share any relevant information we find. If a question arises, I consult with my geneticist in Mexico, and some members offer to check with their doctors. When I find people or communities on social media sharing information related to LCA, I invite them to join the group or follow related pages. Our goal is to provide information and mutual support.

I also created a Facebook group for people with my genetic mutation, similar to the RPE65 group and others. In the *RPGRIP1* group, we communicate closely with Odylia Therapeutics.

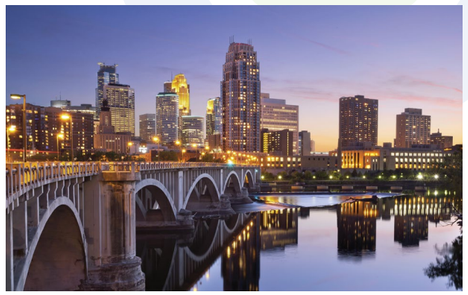
In 2019, I started a civil association in Mexico, equivalent to a United States foundation. My idea was to help and receive support, but Mexico's legal aspect is very complicated. After careful

Two photos above: Angélica Bretón Morán at the 2024 Blind Concert in Mexico

consideration, my family and I decided to dissolve the association and continue supporting the community in other ways.

What are your hopes for the future?
I hope to find better ways to continue supporting the community and that everyone with an LCA diagnosis finds a strong support group. From experience, unity is more powerful than material resources. It is crucial that we not only look out for ourselves but also for others. Although the future is uncertain, that's nothing new to me. I do everything within my power and fully trust that my safety is in God's hands. He knows what I don't, and that's enough to keep moving forward.

If you or someone you know has Leber congenital amaurosis (LCA) due to the *RPGRIP1* gene mutations (LCA6), email info@hopeinfocus.org to join our contact database so we can give you up-to-date happenings with your gene.



HOPE in FOCUS

LCA Family Conference

June 20–21, 2025 • Minneapolis, MN

EARLY BIRD PRICING UNTIL APRIL 1

Registration is open!

Scan the code or visit hopeinfocus.org for more info, registration, and sponsorship.

The Hope in Focus LCA Family Conference will provide information about advances in research, deepen your understanding of various stakeholders' roles in developing treatments, and provide an opportunity for those living with rare inherited retinal disease (IRD) and those involved in developing treatments to learn from each other. The conference offers opportunities to engage in a robust and interactive exchange of knowledge, ideas, and viewpoints as you make new connections in the LCA and IRD communities.

CONNECTING

Patients
Caregivers
Advocates
Industry

EMPOWERING

The community to use its voice in advocacy

ACCELERATING

Research by bringing the patient voice to industry

"Hope in Focus has been such a pillar of support for my family and me, I'm not sure if I'll ever be able to thank them enough. They have watched Gunner grow in every aspect over the years and have empowered me to be the best I can be for him in addition to advocating for families like us in the visually impaired community. I enjoy (and always look forward to) attending Hope in Focus's conference for the personal opportunity to engage with families that have a child/children the same age or older than Gunner so I can learn from them, and share my knowledge and experience with those that have a child/children younger than Gunner. Hope in Focus is a family and I can't imagine this adventure without them."

— Ashlyn Lincoln
Parent of a child
living with LCA

"Atsena's participation in the 2023 LCA Family Conference was a truly special opportunity to connect with the LCA community, and we're excited to support this invaluable event again in 2025. It's essential to bridge research and science with the individuals who experience it daily. Hearing directly from families not only fuels our work but also strengthens our commitment and drives our motivation."

— Kara Fick,
Director of Patient Advocacy
and Medical Affairs,
Atsena Therapeutics

Encouraging Update on MeiraGTx's *AIPL1* Gene Therapy and Other Programs

Ben Shaberman
Vice President,
Science Communications
Foundation Fighting Blindness

The path forward for MeiraGTx's gene therapy for Leber congenital amaurosis 4 (LCA4), which is caused by mutations in *AIPL1*, has not been straightforward for two primary reasons: First, LCA4 is rare, affecting only a few hundred people in the US and less than 10,000 people globally. Second, LCA4 is a severe form of LCA. By about four years of age, most children with LCA4 have lost virtually all their photoreceptors (rods and cones), the cells in the retina that make vision possible. After that age, gene therapy is not a feasible treatment approach.

MeiraGTx, a genetic medicines company in New York and London has manufactured its LCA4 gene therapy under a specials license from the Medicines and Healthcare products Regulatory Agency (MHRA) in the UK. Like many other retinal gene therapies, the treatment is injected underneath the retina and uses a human-engineered adeno-associated virus (AAV) to deliver copies of the therapeutic gene (AIPL1) into remaining photoreceptors.

A total of 11 children received the gene therapy under the specials license and with local ethics approval. All gained visual acuity and showed improvements in functional vision. More about these children in a moment.

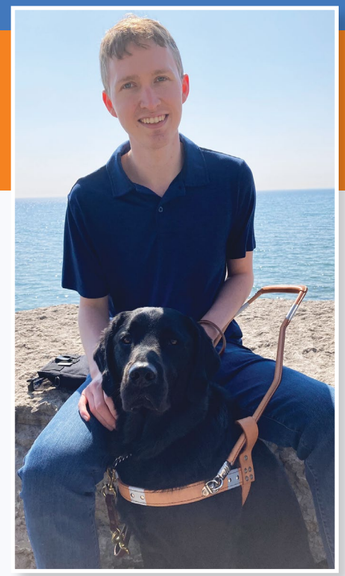
The encouraging news is that the company announced recently it was awarded an Innovative Passport Designation for the gene therapy from the MHRA. The designation can accelerate the time to market and patient access. As a result, MeiraGTx hopes to apply for marketing approval in the UK for its LCA4 gene therapy.

At the annual meeting of the Association for Research in Vision and Ophthalmology (ARVO) in April 2023, I attended a presentation on the *AIPL1* gene therapy, which included videos of the first four children who were treated with the *AIPL1* gene therapy at Great Ormond Street Hospital in London. They ranged in age from 10 months to 2 years and 10 months. These children had very little vision at baseline but were able to identify small objects (e.g., a 1 mm object) after treatment. One child could distinguish different colored pens. I was moved and impressed by the vision improvements.

MeiraGTx also recently reported a regulatory update for all their inherited retinal disease programs. They received Rare Pediatric Disease Designations from the US Food & Drug Administration (FDA) for the following emerging gene therapies: *AIPL1*, *RDH12*, *BBS10* and *GUCY2D*. The designation can enable the company to get priority review for these treatments if and when applications for marketing approval have been submitted to the FDA. MeiraGTx is also seeking partners to further advance these programs as well as its gene therapy programs for achromatopsia caused by mutations in *CNGA3* and *CNGB3*.

Jack McCormick column

ADAPTING TO THRIVE: Navigating New Communities



Jack and his guide dog, Baloo

In the September 2024 Hope in Focus (HIF) newsletter, I wrote about the critical role that finding a community like HIF plays in the lives of people who are visually impaired. In that column, I shared how these communities helped me feel less alone while providing opportunities to learn from others facing similar challenges in a world not designed for the visually impaired.

However, finding and connecting with groups outside the visually impaired population is critical. I've met people who are so immersed in the visually impaired community that they struggle when engaging with those outside of it. I must admit that the challenges associated with interacting with the world beyond this community can sometimes feel overwhelming.

As a child, my parents allowed me to try many different activities, such as swimming and piano lessons, soccer, karate, and more. Participating in these pursuits taught me that with hard work, I could master new things. They also helped me develop my most important skill—the ability to adapt.

Let's discuss why adaptability is vital for people who are visually impaired. Many accessibility, social, and attitudinal barriers impact us. The sooner someone can experience these challenges in a safe environment, like having a piano lesson with an understanding teacher, the better they can adapt to a more challenging situation, such as dealing with a professor who refuses to implement accommodations. I'll expand on this idea by sharing a personal example.

As a teenager, I got really into wrestling. But I never wrestled with another visually impaired person. Instead, wrestling rules have specific accommodations for wrestlers who are visually impaired, requiring that their opponent never break contact with them. My primary training partner eventually became one of the top wrestlers in the country for our weight category. While training with him, I frequently lost, but I won many of the matches that counted!

Because I developed confidence and an ability to adapt early in life, I found the wrestling community, which became my safe place throughout high school. If I hadn't developed resilience through wrestling, I would have struggled with my education and not been as successful in my career. Stepping into these experiences developed resilience and adaptability, which later helped me find an employer that celebrates my capabilities, challenges me to develop my skills, and provides a safe environment to advocate for myself when something isn't accessible.

By sharing a few of my experiences, I hope to encourage parents to find various communities to help their children who are visually impaired to develop belonging and capability. A visual impairment isn't always the most significant stumbling block—sometimes, it is fear of the unknown. If you have a visual impairment, please continue developing your skills and expanding your connections with other communities. You will be surprised by how enriching and developmental it is to widen your circle of relationships and try new experiences.

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.

Walk to Fight Rare Diseases

April 26, 2025 • Hamden, CT

runsignup.com/Race/Register/?raceId=122820

Hope in Focus will join rare disease organizations across Connecticut as we raise awareness and funds for our organizations. Support the effort by walking in person or virtually with us! Be sure to select “Walk for Hope in Focus” when registering.

Hope in Focus • LCA Family Conference

June 20–21, 2025 • Minneapolis, MN

hopeinfocus.org

The Hope in Focus LCA Family Conference will provide information about advances in research, deepen your understanding of various stakeholders’ roles in developing treatments, and provide an opportunity for those living with rare inherited retinal disease (IRD) and those involved in developing treatments to learn from each other. The conference offers opportunities to engage in a robust and interactive exchange of knowledge, ideas, and viewpoints as you make new connections in the LCA and IRD communities.

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. Walks happen throughout the year. Join a VisionWalk in your community! Together, we step closer to fighting blinding 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, Director of Outreach and Development
- 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:
- Spark Therapeutics
- Johnson & Johnson Innovative Medicine
- 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