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

SEEING HOPE | Newsletter

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FROM THE WALL: Pushing Boundaries, Building Confidence

By Katherine L. Kraines, MS

Michelle on a climbing trip with her first guide dog, Ryder

Throughout her 29 years, Michelle Ward Caton has embraced life with unwavering curiosity and gusto. Determined to test her abilities and not be limited by vision loss from Leber congenital amaurosis (LCA), she is, among other things, a competitive adaptive rock climber and teacher of the visually impaired (TVI).

Growing up in North Haven, Connecticut, Michelle's route to an LCA diagnosis took time. Initially, her parents noticed that she wasn't focusing on faces. "Once I was walking, I bumped into a lot of things! But when this was discussed with my pediatrician, he said it was Michelle being Michelle," she said. "When I was three, we moved into a new house, and while running around, I hit my head on the kitchen island overhang. My concerned parents took me to a pediatric ophthalmologist, where my poor eyesight was finally confirmed."

Several years later, the eye doctor detected white blood cells in her eyes and sent her to a Boston eye specialist. By the time they got to Boston, the white cells were gone. However, it wasn't until she did genetic testing during high school that Michelle was diagnosed with LCA8 from a mutation in the *CRB1* gene.

The Journey

Early intervention services started at age three, and by age five, Michelle had a TVI. Refusing to be sidelined, she did everything that her older sister did at the same time. "If she learned to bike ride or swim, I did too!" Michelle said. "We were only a year apart, and it was almost like being twins. But determined to keep up, I did many daring things for a blind person."

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From the Director

Courtney Coates

Dear Friends,

I'm excited to announce our upcoming Party for Sight summer fundraising campaign! This event is more than a fundraiser—it celebrates hope and progress in the journey toward treatments for inherited retinal diseases (IRDs). Your participation in the campaign will make a lasting impact, helping to drive awareness of rare retinal disease and support for Hope in Focus.

I'm also excited to see you at our 2025 LCA Family Conference, an event designed to bring together individuals and families affected by Leber congenital amaurosis (LCA). This will be a fantastic opportunity to connect, share stories, and learn from experts in the field.

I've been on the road recently attending conferences, and I'm happy to share that so much progress has been made toward treatments for IRDs. This fact reinforces how important it is to continue to engage and support our patient community, something we cannot do without your help.

Thank you for your continued support. I look forward to Partying for Sight with you this summer!

Warmly,

Courtney Coates

Courtney Coates

FROM THE WALL: Pushing Boundaries, Building Confidence

Continued from page 1

Her resolve to play softball in elementary school was dashed, she said, "When my dad took me outside and tossed a few softballs at me until I realized I couldn't play. When you're a kid, you think you can do anything. Sometimes, you have to realize you can't."

Michelle's parents exposed her to various activities, including the piano, flute, and bass guitar. Later, she participated in gymnastics and was a ballroom dancer throughout high school. "I did Brazilian Jiu Jitsu and then Muay Thai, a form of kickboxing, which my dad practiced with me, allowing me to get the rhythm of the sport and have incredible workouts," she said.

In college, Michelle tackled the gym and weightlifting, and when she was at UMass Boston working on her TVI degree, she met a friend who did rock climbing. "I began climbing with an adaptive climbing group that participated in competitions," she said. "I went to nationals and placed second, got on the United States team, and went to the international competition in France," she said.

Rock Climbing Lessons

Michelle and her husband, Jason, continue to climb and discuss competing again. "It's important to go out of your comfort zone. I still have some vision, and being outside and up high with a view is incredible," she said. "When I'm climbing in the gym and hit the spot where I've fallen many times and finally figure out the right move and body positioning, it's a small victory. It's great getting to the top of the wall, whether I can see it or not. The top of the wall isn't necessarily the prize. It's the fact that you did it that builds confidence."

Children with visual impairments often find body awareness and balance difficult, but these can also be tough for adults. Sometimes, when Jason tells Michelle how to move on the climbing wall, getting her correct body part to the right location is challenging. "Jason might say to move my right foot three feet, and I move my left foot one foot," she said.

Helping children with body awareness is something Michelle focused on teaching in her preschool classes during TVI training. "I'd set up obstacle courses for the children to climb over, under, or step through or over," she said. "I think climbing and Jiu Jitsu are some of the best sports for kids with vision impairments because they help build body awareness."

Vision Changes

When Michelle was about 26, her vision dramatically worsened. "I abruptly lost acuity, and my nystagmus got much worse. There was no time to adjust," she said. "I've always had some nystagmus but could focus through it; now my vision always vibrates."

As a TVI, Michelle teaches braille, but she also uses it to access all of the appliances in her home. She does most of her visual work in the morning because the nystagmus exhausts her by evening. "You have to adapt your scenario to your specific needs. It feels like I'm constantly adjusting. I keep puzzling things together to see what works," she said. "This is the reality for everyone with LCA; each situation is different, and you must finesse things until something works."

The Right People

Dating during college helped Michelle discern that "it's the people that don't care about my vision that make the best friends. I'd meet someone, and when they found out about my vision impairment, they would shift from 'You're cute!' to 'You're a science project.' They'd ask me how many fingers they held up and what I could see. I got all of the stereotypical questions."

Jason and Michelle, with guide dog Ryder, at Arches National Park

Michelle at the 2019 national adaptive climbing competition

Michelle loves that her husband doesn't care that she is blind. "Jason does things for me that I don't even notice. I'll be looking for something, and he will grab it, hand it to me, and walk away without commenting. Or he will get out of the car and offer me his arm. We never talked about my limitations—he just got it!"

She said finding supportive people who don't coddle is essential. "Jason encourages me to learn new things that a friend might say are dangerous. Instead, he teaches me how to do it. He doesn't make assumptions about what's possible for me."

A sense of humor is vital for navigating a degenerative condition; Michelle explained, "I trip and fall and knock things over all the time! My visual decline in the last couple of years has been very tough, which makes it essential to find someone who laughs with you. Being morose doesn't work!"

Family Interactions

Reflecting on growing up with LCA, she said, "My parents always treated me like my sister. Being blind was a characteristic that only meant I did things differently. My sister didn't acknowledge my being blind for a long time. I don't think she fully realized what it meant. Instead, she would stare at me and say, 'Just do it,' which is what a big sister does."

"My parents never wallowed in my diagnosis, and we had many real-life conversations about my vision loss. But my dad struggled because he knew my vision would get worse, and I would face lifelong challenges." Michelle said. "In high school, we met Robin Clark, a teacher passionate about ensuring kids had the independent living skills to live successfully. We implemented creative strategies, worked on things at home, and discussed what would help."

TVI Responsibilities

As a TVI, Michelle travels between schools in New Hampshire, helping elementary through high school-aged students based on their individualized education plans (IEPs). "I love working with the teams that work daily with the kids to support their vision in class," she said. "Life with LCA is like rock climbing. Starting out, you have no idea what you are doing, but you feel your way along, and eventually, you're competing!"

If you or someone you know has Leber congenital amaurosis (LCA) due to the *CRB1* gene mutations (LCA8), email **info@hopeinfocus.org** to join our contact database so we can give you up-to-date happenings with your gene.

HOPE in FOCUS

PARTY FOR SIGHT

May 2025–September 7, 2025

PARTY WITH PURPOSE!

Join Hope in Focus for our summer-long fundraising campaign, Party for Sight, a fun and meaningful way to raise money for Hope in Focus and drive awareness of Leber congenital amaurosis (LCA) and other rare inherited retinal diseases (IRDs).

IT'S SO EASY! Get together with friends and family, share some rare disease facts and ask your guests to make a donation. And, of course, have fun!

NEED INSPIRATION? Do what makes you happy! Party for Sight is an opportunity to gather your friends and do something you enjoy while also helping our nonprofit.

- **Backyard Picnic:** A casual gathering with good food and friends.
- **Cocktail Dinner:** Share your signature drink and appetizers.
- **Paddle for Sight:** Organize a group kayaking trip or another outdoor adventure.
- **Paint & Sip:** Get creative while raising funds!
- **Game Night, Birthday, Trivia Night:** Make it your own!

Ready to make an impact? Text "PARTYFORSIGHT" to 71777 or scan this code for more info.

Thank You

FOR YOUR SUPPORT IN 2024!

INDIVIDUALS

Elizabeth Adrian & Dr. George Adrian
Melissa & Michael Allen
Steven Alper
Matthew Amaro
Kym Apicelli & John Spinnato
Cari & William Barnes
Jim Bates
Christine Bohmann
June & Thomas Bordner
Beth & Andrew Borysewicz
Paul Bourguignon
Tracy Bourguignon
Marcie & Howard Brensilver
Patricia Brigham
Stephanie & Kyle Brown
Kim Cardinal
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TMG Plumbing & Disaster Solutions
Town Fair Tire Foundation
Trident Revenue Management
Valenti Auto Mall
The Whaler's Inn

BlueRock Photoreceptor Replacement Therapy Moving into a Clinical Trial

Ben Shaberman
Vice President,
Science Communications
Foundation Fighting Blindness

FOUNDATION FIGHTING BLINDNESS

The development of gene therapies for inherited retinal diseases (IRDs) took off when young adults and children showed significant vision improvements in an early clinical trial for what would become LUXTURNA® for LCA2 (*RPE65* mutations). That was in 2008. LUXTURNA became the first FDA-approved IRD gene therapy in 2017. Thanks to that success, dozens of gene therapy clinical trials are underway. Some target specific genes. Others are gene-agnostic, designed to preserve photoreceptors or harness non-light-sensing cells in the retina, an approach called optogenetics.

Cell-based therapies for IRDs have not advanced so quickly, with just a few clinical trials being launched. Gene therapies, which use human-engineered viruses to deliver the therapeutic gene, are not easy to develop or administer. But cell therapies, especially those for replacing lost photoreceptors, present additional challenges that have been difficult to overcome. These include determining the source and manufacturing of the cells, promoting their survival after transplantation, and enabling their integration into the host retina.

The launch of a Phase 1/2 clinical trial for OpCT-001, an emerging photoreceptor replacement therapy from BlueRock Therapeutics, is a big step forward for the IRD cell therapy field. The trial will initially enroll people with IRDs such as retinitis pigmentosa and cone-rod dystrophy. But the approach could also be relevant for forms of LCA that primarily affect photoreceptors. OpCT-001 is comprised of photoreceptor progenitors—photoreceptors that haven't fully matured. Researchers believe that progenitors have the best chance of integrating and surviving once they are transplanted and mature. The progenitors are developed from induced pluripotent stem cells (iPSC). To produce iPSCs, investigators take a small blood or skin sample from an adult human donor. The cells are then genetically tweaked to revert to a stem-cell-like state. As stem cells, they can be coaxed to develop into virtually any cell type in the body, including photoreceptors. Furthermore, billions of cells (many therapy doses) can be produced from the cell sample. The study will assess several dose levels of the therapy and is expected to enroll participants in sites across the U.S.

BlueRock Therapeutics is a wholly owned subsidiary of Bayer AG. The company licensed OpCT-001 from FUJIFILM and Opsis Therapeutics, a company co-founded by David Gamm, MD, PhD, a world-renowned retinal cell therapy pioneer at the University of Wisconsin-Madison. The Foundation Fighting Blindness provided significant funding over several years to Dr. Gamm and his team for the development of retinal and photoreceptor cell therapies derived from iPSC.

One should never get too excited about any emerging therapy in an early-stage clinical trial, especially for something as cutting-edge as a photoreceptor progenitor treatment. But if there is one scientist on the planet who can get photoreceptor replacement to work, it is Dr. Gamm.

Stay tuned.

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

We take great care to use every dollar we raise to advance our mission.

2024

In 2024, Hope in Focus reflected on its past decade as an organization and devoted significant time to planning for the future. We heard clearly from the LCA community that our grassroots outreach, family support, and educational activities are a key focus for them, and one which we are uniquely positioned to provide.

Considering these needs, our 2024 research contribution of $21,000 to the Foundation Fighting Blindness, earmarked for "LCA therapies," was considerably lower than in previous years. This contribution included 100 percent of the dollars raised that were designated for research, plus an additional 2:1 match from our undesignated net assets.

The 2024 contribution reflects the Board of Directors' recognition of the importance of continuing to support research, while acknowledging the need to expand outreach and advocacy programs requested by the LCA community.

As we celebrated our 10th anniversary at Dinner in the Dark, we are pleased to report this was our highest-grossing event to date, raising over $225,000. Despite being a crucial part of our program support, corporate sponsorships remained unchanged from 2023.

Looking ahead...

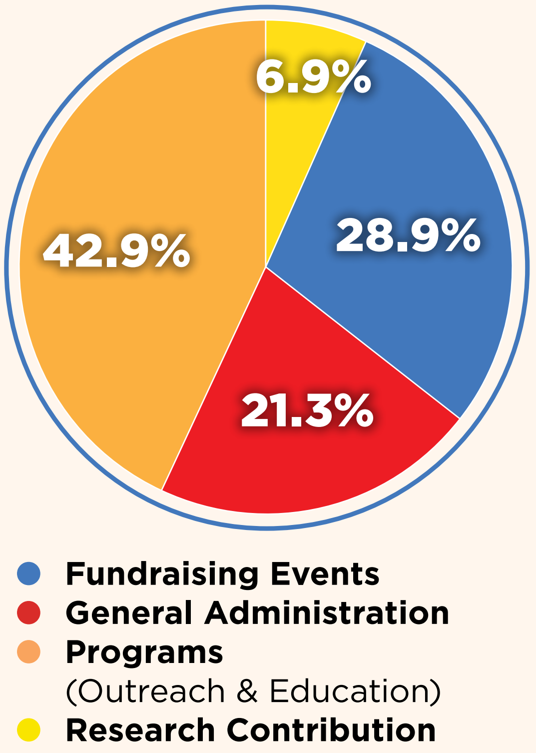
Hope in Focus operates in a constantly changing environment. Treatment research continues to advance, our global community grows with improved access to genetic testing and healthcare information, and patient needs shift from seeking basic disease information to evaluating potential treatment modalities and considering clinical trials.

To keep pace with these changes, we plan to grow our Hope in Focus team while actively pursuing opportunities to diversify our funding sources and expand our donor base.

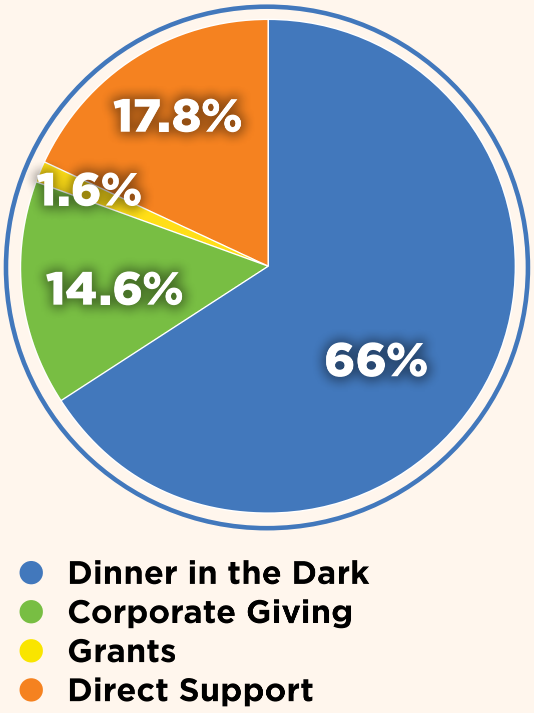
We hope you will support our work, enabling Hope in Focus to continue advancing research and ensuring that the patient voice is heard.

For more detailed information and to view our past Form 990s, visit our profile on GuideStar.

EXPENSES



INCOME



Events

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

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.

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.

HOPE in FOCUS

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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

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