So, Where Were You?

I remember exactly where I was when we received Sofia’s clinical diagnosis of LCA. And I remember exactly where I was when the lightning bolt struck and I realized—really realized—she was considered legally blind and would at some point in the future lose her sight completely.

These two events were months apart—the real “sinking” in happened in a room full of teachers discussing Sofia’s Braille studies and staring sideways at me. Suddenly I was the mom who knew her daughter was blind, had pushed for this meeting, and yet was freaking out and wondering why her daughter needed Braille instruction. Clearly, one of my finer moments.

Of course I listened to the professionals, asked questions and Googled LCA a thousand different ways looking for that one article or post that I could understand, relate to and that might even offer some clear answers for what the future would hold. Despite it all, like many other families in the LCA community, we didn’t know anyone else who had a rare retinal disease, or even any other blind children.

Even eight years later, I still have those struck-by-lightning moments.

continued on page 3
Finding treatments for the 27+ genetic forms of LCA is no doubt a challenging problem, but promising advances are being made on several fronts, especially in gene-therapy development.

Spark Therapeutics’ RPE65 (LCA2) gene therapy, now meeting name LUXTURNATM, is poised to become the first gene therapy for inherited or acquired conditions to receive FDA approval, thanks to its safety and the impressive vision restoration it delivered to children and young adults with severe vision loss in clinical trials.

Spark has filed its biologic license application (BLA) for LUXTURNATM with the project date for an approval decision of January 12, 2018. Spark is also seeking regulatory approval for the treatment in Europe.

FDA approval for LUXTURNATM would provide strong affirmation for the biotech industry that gene therapy is a viable approach to treating LCA, a broad spectrum of inherited retinal diseases, and other genetic conditions.

Several other LCA therapies are advancing through the development pipeline. Here are overviews of some of those efforts:

**Clinical Trial Authorized in the U.S. for LCA10 (CEP290) Treatment**

ProQR, a biotechnology company in the Netherlands, has received authorization from the FDA to start a Phase I/II clinical trial for its therapy known as QR-108, which is being developed for LCA10. QR-108 targets the specific mutation in CEP290 gene that causes the disease.

**Emerging Gene Therapy for LCA5 (RPRG1) Mutations**

A team of University of Pennsylvania researchers has identified a canine model of LCA caused by mutations in the gene IQGB1, which encodes the protein NPHS5. In humans, the condition causes early and severe vision loss from infancy and in some cases, kidney dysfunction. The researchers found that, unlike the mouse, the retinal degeneration is remarkably similar to that in humans with NPHS5 mutations, though dogs don’t have the renal dysfunction. Using the model, the researchers are now testing gene therapy approaches to either slow or halt vision loss, or possibly improve vision, in humans. They’ve already had some success with gene therapy in canines.

Researchers Pursue Gene Therapy for LCA Caused by IQGB1 Mutations

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**Projects supported by the Foundation Fighting Blindness**
I noticed a light that I couldn’t target those who are different. I was bullied a lot—vision impairments. I went to school and made many of its own memories with my family. When I was diagnosed with Leber congenital amaurosis in 2012, about the time their daughter, Annie, was being mainstreamed into the local school. In 2009, doctors diagnosed Annie at 3 months old with LCA-CEP290. After being inspired at the Foundation for Retinal Research conferences, the Cohanes decided to get involved. Their objectives were: to help fund LCA research; to raise awareness for Annie in her community, as well as others who are blind; to bring people together and to have fun. They decided on a simple idea of a “backyard tailgate” with open bar, snacks, sweets and a rolling live band and a suggested donation at the door. Since 2012, they have raised over $130,000 and their most recent party had over 350 attendees under the stars. This year, they hope to continue their success with their “Back to Basics” party in Norwell, Mass., on Saturday, Sept. 23. In the past, the Cohanes gave the funds to the Foundation for Retinal Research. Now the funds will go to the Foundation for Fighting Blindness since the two organizations merged. Congratulations to the Cohanes for hosting such fun and fruitful parties!

Dear Diary:
I wish I could see more stars

August 22, 2017
Dear Diary,

I like the stars, even though I can’t see them. I was excited for the eclipse. Everyone was talking about it, and since it’s summer and mom works from home, she would be there to watch it together. Dad got a brownie box and a cereal box and made some sort of contraption for us to safely watch the eclipse. I joked that it was funny that the sun was in the box. Also that I might damage my eyes more, since I’m going to be totally blind someday anyway.

Once the eclipse started, we all hung out on the porch and looked into our boxes. I could see the dot inside the box. I couldn’t tell that sun was being covered though. Only that the light was getting dimmer. Mom and Dad kept asking me what I could see, and I kept telling them I could see it. I think that made them happy.

When I look up in the night sky I just see darkness. Sometimes I can see the moon. Sometimes I can see some really bright stars. Once, I was excited to see the moon huge and bright but Mom told me that it was just a street lamp and I was looking in the wrong direction. But the other times I know that I saw moon. I wish I could see more stars. Someday I hope to see all the stars, but I guess I probably won’t. I hope I’m wrong.

Sincerely,
A Stargazer with LCA
Age 14

Jenny and Kevin Cohane began raising funds for Leber congenital amaurosis in 2012, about the time their daughter, Annie, was being mainstreamed into the local school. In 2009, doctors diagnosed Annie at 3 months old with LCA-CEP290. After being inspired at the Foundation for Retinal Research conferences, the Cohanes decided to get involved. Their objectives were: to help fund LCA research; to raise awareness for Annie in her community, as well as others who are blind; to bring people together and to have fun. They decided on a simple idea of a “backyard tailgate” with open bar, snacks, sweets and a rolling live band and a suggested donation at the door. Since 2012, they have raised over $130,000 and their most recent party had over 350 attendees under the stars. This year, they hope to continue their success with their “Back to Basics” party in Norwell, Mass., on Saturday, Sept. 23. In the past, the Cohanes gave the funds to the Foundation for Retinal Research. Now the funds will go to the Foundation for Fighting Blindness since the two organizations merged. Congratulations to the Cohanes for hosting such fun and fruitful parties!

Have an update, event or a shout-out to share? We want to hear from you! Send us your news at rosanne@sofiaaseeshope.org

Jack McCormick is a business student at Canada’s Wilfrid Laurier University, and a guide dog user who lives with Leber congenital amaurosis, an inherited retinal disease. He is an active accessibility advocate who dreams of a future where society no longer defines people by single characteristics. Upon graduation, Jack hopes to start work in the field of Human Resources and continue to be an accessibility advocate.

You can read his blog here: jackdamccormick.wordpress.com.

“It took well over a year, countless procedures and eventually genetic testing to find out that the doctors were wrong.”
The Role of the Patient Voice in Research

By Rosanne Smyle

Extraordinary and exciting developments are happening in LCA research, as Spark Therapeutics’ gene therapy for an LCA gene mutation is under review by the Food and Drug Administration, in the hopes of becoming the first gene therapy for a genetic disease in the United States.

The therapy under FDA review would be for patients with vision loss due to confirmed biallelic RPE65 mutation-associated retinal dystrophy, according to the FDA’s Department of Health and Human Services notice.

In September, the FDA announced the public meeting of the Cellular, Tissue, and Gene Therapies Advisory Committee (CTGTAC) on Oct. 12 in Silver Spring, MD. The committee will discuss and make recommendations on the safety and effectiveness of an application for voretigene neparvovec, which has the proposed trade name of LUXTURNAN™, submitted by Spark Therapeutics, headquartered in Philadelphia, according to the notice.

The role of the patient community is important long before a potential treatment gets to the FDA, and patients and advocacy groups play a pivotal role in moving research to fruition.

Patient input is especially important in what can be a long and arduous process, hampered by research setbacks and by the business of research when companies are bought and sold or when personnel move from one company to another. Connecting researchers’ work to patients’ needs helps inform the research and also motivates teams as they continue their studies.

Researchers work at sustaining two-way conversations between patients and companies by building and growing relationships. This happens through advocacy organizations and through formal patient advisory boards to gleam answers to researchers’ anticipated issues.

Increased awareness of rare inherited retinal diseases and recent FDA approval of the first gene therapy in the U.S. for a form of non-genetic leukemia send a positive signal to the rest of the biotech industry, encouraging more gene therapy research.

The rare inherited retinal disease community should be encouraged and ready to participate in patient registries, advocacy organizations, patient advisory panels and other awareness opportunities where their voices will continue to support and inform research to cure blindness.

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Trust me when I tell you to grab a tissue. Or roll down your sleeves and get ready to wipe. Me? My eyes are welling up, just a little type. Recently, I witnessed a miracle.

I’ll give you a little background while you look for a Kleenex. Twenty years ago, at the age of 8 weeks, my nephew Alan was diagnosed with Leber congenital amaurosis (LCA), a rare disease that limits retinal development.

Holding her infant son, my sister Betsy and her husband David (listened in disbelief as the retinal specialist explained that Alan would be visually impaired at best, and fully blind at worst. The doctor informed them there was no cure, no treatment, and no adaptive device to correct their son’s condition.

Like his sighted peers, Alan attended public school. His mobility instructor taught him to navigate familiar parts of his world with his white cane. By sixth grade, he could walk to school by himself. Now a junior at Beloit College, he’s doing just fine. That’s not the miracle.

Not to dismiss Alan’s role in his accomplishments, but his successes have been in some part reliant on a team of people. Still, there are limits to his independence. Sooner, or later, here comes the miracle.

Last week, Betsy called me. “Get outside now. Alan is walking to the train station. By himself. He’s near the library. Run!”

I ran. Turning the corner, I saw my nephew, cane in hand, walking a route new to him. Betsy trailed silently, about 10 feet behind.

“Hey Aunt Sally. Is that you?”

Cue the tears. Remember, Alan is blind. He has never seen me coming his way. For 20 years, we’ve all come up to him and touched him, or spoken to him, or hugged him, letting him know we were there. For the first time, he ‘saw’ me coming.

Alan was wearing an adaptive technology, and it was changing his life in front of my tearing eyes.

The technology is called Aira (eye-rah). According to their website, “Aira’s platform works on a wearable device similar to Google glass, that can be paired with a smart phone. The tiny camera mounted on the device [cool sunglasses] provides instant feedback to a trained Aira agent who can safely guide [a wearer] in any activity.”

Alan’s Aira glasses had arrived the day before. The device is free, and the glasses are free. Like a cell phone contract, users pay a monthly service fee based on the minutes they use. When it’s on, an agent sees a split screen. On one side is a GPS view, to map out the exact location of the route guidance. On the other screen, the agent sees the lens view from the user’s camera. Simple, yet genius.

I fell in step with Betsy, following Alan’s lead for the first time. He made it easily to the train station, then home via a different route. Since then, he’s walked to a local bagel shop and “read” menus. I asked Alan, “What do you like best about this?”

“I like walking somewhere that I’ve never walked to before, by myself, without learning the route. I like reading menus. I like finding people. I found you!”

I turned to my sister. “Well? What are you thinking?”

“It’s totally selfish. I want to be his eyes.”

“Don’t you think you’ve been his eyes?”

“Yeah, I guess. And now I’m turning it over to somebody else.”

There wasn’t a dry eye between us.

Tell Us Your Story

Independence Day

By Sally Higginson

“For 20 years, we’ve all come up to him and touched him, or spoken to him, or hugged him, letting him know we were there. For the first time, he ‘saw’ me coming.”

“I like walking somewhere that I’ve never walked to before, by myself, without learning the route. I like reading menus. I like finding people. I found you!”

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Events

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

Sofia Sees Hope

Dinner in the Dark
October 14 • 6:00 PM–11:00 PM
Mystic, CT
sofiasees.org/getinvolved/dinner-in-the-dark

Sofia Sees Hope’s primary fundraiser for the year, this event helps fund research to cure blindness caused by LCA, provide support for genetic testing and drive awareness, education and connections for LCA and IRD families. Be prepared for a unique menu, fine wines and a lively sensory adventure!

National Organization for Rare Disorders (NORD)

Breakthrough Summit
October 16 8:00 AM – October 17 @ 5:00 PM
Washington, D.C.
rarediseases.org/summit-overview

National Organization for Rare Disorders (NORD) annual Breakthrough Summit—information about advocacy, meetings for members, potential collaborators.

Foundation Fighting Blindness

Vision Seminar Series
Nashville: Fall 2017
Los Angeles: Winter 2018
San Antonio-Austin: Jan. 27, 2018
Tampa, FL: Winter 2018
blindness.org/conferences

The Foundation Fighting Blindness is proud to enter the ninth year of our successful Vision Seminar Series! With so many advances in retinal disease research, you should not miss this opportunity to learn more about the latest research advancements, treatment options and clinical trials.

Foundation Fighting Blindness

VISIONS 2018 National Conference
June 21–23, 2018 • San Diego, CA
blindness.org/conferences

Wonderful weather and a myriad of family friendly activities! Hear the latest research and treatment advances. Attend sessions on thriving despite vision loss, and enjoy social and community-building opportunities. Conference registration will open January 2018.

What do you want to learn at the VISIONS conference? Send your input and ideas to info@sofiaseeshope.org

Events