SOFIA SEES HOPE

Supporting the LCA and rare retinal disease community.
We see a cure for blindness.

SEEING HOPE | Newsletter

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From the Founder

Twelve years ago, my daughter Sofia was clinically diagnosed with Leber congenital amaurosis (LCA). Seven years after that, she received her genetic diagnosis, IQCB1/NPHP5. At that time, there were 20 genes associated with LCA. Today there are nearly 30.

Sofia Priebe & Laura Manfre

Of course I have reacted to all of this as a mother. I want a cure for my daughter. I want her to realize her dream of seeing the stars in the night sky. I want her to see the leaves on the trees and the shells on the beach.

But I also was struck by the need to do more. As I navigated my way through the world of inherited rare diseases, and more specifically LCA, I discovered there are others like me feeling alone, with many questions and much confusion. I knew I needed to help more than my daughter.

And so in 2014, I founded Sofia Sees Hope with my husband, Chuck, and the help of friends and family. Over the last four years it has evolved from its infancy to what I now consider its adolescence. Through the support of sponsors and individuals, networking and education, we have experienced a breakthrough year in 2017.

Last year, Sofia Sees Hope launched a new website designed to be a hub

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Living with LCA: Enzo's Story

By Rosanne Smyle

Enzo was born in 2014 in Lausanne, Switzerland, with a clubfoot. He immediately received physical therapy, then a plaster cast for three months and one minor surgery.

His parents — Laura Steinbusch, a post-doctoral fellow researching the neuroscience of diabetes, and Merlijn Meens, a scientist investigating cardiovascular function — felt confident as they were reassured through medical literature and by doctors that in time their son's left foot would be fine.

But there was more to come. At 3 months, as Enzo's therapy for his foot progressed, something seemed wrong with his vision and his ability to focus. Doctors diagnosed him with nystagmus, a condition in which Enzo's eyes involuntarily moved side to side.

An MRI showed normal brain development and then doctors did an electroretinogram (ERG), placing electrodes on his eyes to measure the electric response of their light-sensitive cells. Enzo also underwent Visual Evoked Potential (VEP) testing, a non-invasive exam that measures his entire vision system.

Doctors fitted Enzo with glasses at 6 months and said the ERG and VEP results needed further study.

Enzo began rubbing his eyes, prompting people to ask Laura whether her son was tired or shy. A few months later, doctors diagnosed 10-month-old Enzo with Leber congenital amaurosis.

"While walking home from this last doctor's appointment, we were in shock, but after a while we realized that Enzo had not changed. He was still our cheerful son that likes to sing and cuddle," Laura wrote in her blog for the Eye Association of the Netherlands.

"We wondered how we could raise Enzo as normal as possible and how we could help him discover the world. The solution turned out to be simple: We will not despair and (will) come up

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Tell Us Your Story

Allison Galloway

Would you like to tell us your story? Email elissa@sofiaseeshope.org

Somehow I made it through 37 years of life without ever meeting a blind person.

Then my 3-year-old son was diagnosed with Leber congenital amaurosis (LCA), which will cause him to go blind in his teens or twenties. My world was set into a whirlwind of grief, confusion and anger.

This disease caused my son's body to lack the ability to create a simple enzyme called retinal dehydrogenase, which assists in the cleaning and health of the rods and cones in his eyes. It is an autosomal recessive gene defect (RDH12) that appears when each parent has a mutation on the same gene in the same place. There is a 25 percent chance that each child they create will have this disease. The odds were against us, I suppose.

A year and a half later, we learned the odds were even worse than we thought. Our son was 5 and our daughter was 3. One day I noticed my daughter's eye started to bounce from side to side. I immediately knew what it meant. A nystagmus means that the eye is weak and sick. I didn't need the gene results to know that my beautiful little girl had LCA, too.

What fuels my determination to speak out for all those parents dealing with a similar heart-sinking diagnosis occurred at our eye doctor, of all places. The doctor handed me my child's diagnosis on a piece of paper. She told me to schedule a procedure under anesthesia for my son with no explanation of why.

I Googled what was written on that paper. The first word I saw was "blind" and the second phrase that stood out was "no cure." That was all I had. I knew then that I quickly had to learn all I could. The problem is that so many families get the same kind of "rare disease news" and have no idea what to do. They trust their doctors to be the knowledgeable ones. If a doctor says there is no cure, well then there is no cure, right? Nothing left to do but learn how to accept it, scared and alone.

The lack of assistance given to parents is astounding. If I had not had the financial means, the education, and the network, I would have taken the advice of my first doctor and just followed up "if I wanted to."

Knowing your gene is a critical step. The ID YOUR IRD gene testing initiative ends 1/31/18. www.idyourird.com.

Learn more: www.blindness.org/genetic-testing.

As time has gone on I have met so many amazing parents, families, teachers and researchers who are determined to cure many of these rare diseases. Social media has opened so many doors. I was lucky to find a group of 18 families whose children all have the same gene defect as mine. Together we created a network of motivated parents who have formed a 501(c)(3) named RDH12.org to raise money and work with scientists around the world to fund research.

There are amazing things out there like the website, Myretinatracker.org, which was created to generate a database of everyone with inherited retinal disease so scientists can easily access a population to use for clinical trials. Everyone should be on it if they have a genetic disorder.

Genetic testing is free in many instances and all it requires is a simple blood test. Knowing the gene is a critical step toward getting treatment or toward helping advance the research that will lead to cures.

This rare disease is what comes between my children and the world. It limits them. Rare has flipped our world upside down and forever changed me. It keeps me searching for answers and fighting for all those families out there. We know our love is what keeps us going on the days that feel impossible. Love is what will keep us moving on the days when the news at appointments is less than positive. We never give up on our children, and we will never stop fighting for their well-being. All we want is for our children to not have to fight so hard to simply be.

Allison Galloway lives in Westminster, Colorado, with her husband, Michael, and their two children, 6-year-old Logan and 4-year-old Zoe.

You can read her full essay at sofiaseeshope.org/allison-galloway.

Enzo's Story

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with creative solutions so he can participate in everything that this world has to offer."

That Christmas, Enzo's parents designed their holiday in a way that greatly fostered their son's vocabulary, sensory and motor skills, exposing him to chickens on a farm, rocks and water at the beach and a mountain climb with a summit lunch of pancakes and omelets.

Also, for the first time he said, "Dad," "Mom" and a version of his own name, "Echoo."

Then, at 18 months, Enzo received the genetic diagnosis of LCA10-CEP290. LCA10 is a severe retinal dystrophy caused by mutations in the CEP290 gene.

Laura interviewed doctors about research on LCA10-CEP290, becoming an intense advocate and learning myriad facets of the genetics behind Enzo's disease and the stakeholders at work to find a treatment.

With global health organizations headquartered in the Lake Geneva region, Laura attended conferences, including a recent policy event about "the right to health" organized by Rare Disease International.

She learned that researchers in the International Rare Diseases Research Consortium are working toward their goal of all people living with rare disease to receive diagnosis, care and therapy within a year "of coming to subspecialty medical attention."

"Big visions and great goals," Laura wrote in her blog for wonderbaby.org. "It made me hopeful for a future with inclusive education for all our children, job possibilities for all our children, a quick diagnosis for new patients and maybe a treatment for improved vision."

At home, Laura and Merlijn initially did not have access to many Braille and tactile books for Enzo so they created their own. For instance,

they brought to life a hedgehog picture book by fashioning spines with nails and duct tape.

Laura now is working on a multilingual children's songbook with songs in English, French, Dutch and German. She got the idea because Enzo easily learned new words in French or English through songs, especially when he already knew the song in Dutch. Two Dutch foundations said they want to help pay for part of the project, but Laura is still searching for funding and she is reaching out to ask whether anyone knows of groups she can contact for help in the United States, United Kingdom, Canada or Australia.

Enzo is now 3. He loves playing with other children at nursery school and he is learning to be more independent. Also, doctors successfully treated his foot, with Enzo's mom adding, "It was probably harder on us than on him."

In September, he and his parents moved from Switzerland to their homeland, the Netherlands.

"Enzo is doing very well," Laura said. "He loves listening to music, singing, reading books, everything that has to do with movement. He adores taking the bus, the train, the metro, the cable car, a bike ride…

"One of his favorite sentences nowadays is 'What is that?' (something he heard) or 'Who is that?' (someone talking), meaning that he is really curious."

Would you like to share your story with Sofia Sees Hope? Email rosanne@sofiaseeshope.org. Read more "Living with LCA" stories on our website: sofiaseeshope.org/resources/blog.

From the Founder

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of information, education and connections for the LCA community. We renewed our focus and energy on advocacy for the IRD community.

We traveled across the country to conferences not just to learn more about what we can do, but also to connect people who need to find help and support. We testified before the federal Food and Drug Administration's Advisory Committee as Spark Therapeutics presented their research and findings on LUXTURNA™. We created this quarterly newsletter that includes information on research, advocacy, events and stories from families living with LCA. At year's end, our 4th annual Dinner in the Dark hosted nearly 400 people and raised almost $200,000.

You need to crawl before you walk and walk before you run. Sofia Sees Hope is in the starting blocks and ready to go ahead at full speed. We have plans for 2018 and beyond that we are confident will move our community forward in the realms of research, education, advocacy and support. We look forward to sharing the journey with you.

Laura

In January 2014, Laura Manfre founded Sofia Sees Hope. The nonprofit funds the development of cures not just for her daughter Sofia's LCA gene, but also supports diagnosis, treatment and cures for all children and adults suffering from blindness caused by any of the 27 genes related to LCA.

Many Genes, One Community

Leber congenital amaurosis is a rare inherited retinal disease characterized by severe vision loss at birth. While some children are born with little or no vision, others may have significant vision loss in the first few years of life, stable vision for a period of time, and then eventually complete vision loss as the retina deteriorates into total blindness.

We often hear of only a handful of gene mutations caused by LCA. There are currently an estimated 27 validated LCA genes according to the Retinal Information Network.

AIPL1
CABP4
CEP290
CCT2
CTNNA1
CYP4V2
CLUAP1
CRB1
CRX
DTHD1
GDF6
GUCY2D
IFT140
IQCB1
KCNJ13
LCA5
LRAT
NMNAT1
PRPH2
RD3
RDH12
RPE65
RPGRIP1
SPATA7
TULP1
IMPDH1
OTX2

Ben Shaberman
Director, Science Communications
Foundation Fighting Blindness

A Cut-and-Paste Approach to Saving and Restoring Vision

FOUNDATION FIGHTING BLINDNESS

By Ben Shaberman, Director, Science Communications Foundation Fighting Blindness

Gene editing is a promising technique for treating inherited retinal diseases (IRDs) such as Leber congenital amaurosis (LCA). A technology called CRISPR/Cas9 is at the forefront of researchers' gene-editing development efforts. CRISPR/Cas9 moved into the public spotlight in 2015 when a team of Chinese investigators used it in a lab experiment to genetically edit human embryos that carried a mutation for a fatal blood disorder.

The editing of human embryos with CRISPR/Cas9 is raising all sorts of ethical questions. It could be a "design tool" for customizing the features (e.g., eye and hair color) of an unborn child or enhancing his or her athletic potential.

However, for children and adults already born with IRDs such as LCA, CRISPR/Cas9 is showing strong potential as a safe and elegant approach for saving and restoring vision.

Gene editing vs. gene therapy

While both techniques address genetic diseases, gene editing and gene therapy are different. Gene therapy, which has been in clinical trials for IRDs for more than a decade, involves delivery of a whole new gene to replace the mutated gene. Spark Therapeutics' RPE65 gene therapy, which is poised to become the first FDA-approved gene therapy for the eye or an inherited condition, is "whole gene" replacement. (This technique is also referred to as gene augmentation or compensation, because the old gene isn't removed.)

In contrast, CRISPR/Cas9 is a gene "cut-and-paste" technology. CRISPR (Clustered Regularly Interspaced Short Palindromic Repeats) finds the mutation in the gene. Cas9 works like a molecular scissors to cut out the mutation. In some cases, a healthy piece of DNA is also inserted to fix the genetic defect.

Think of it this way: Your car won't start. You have two options: Get another car or fix the part that's causing the problem. Getting another car is like gene replacement. Getting a new part to fix the problem — like a battery or fuel pump — is like CRISPR/Cas9.

Pros and cons

Both CRISPR/Cas9 and gene replacement have their advantages. A plus for CRISPR/Cas9 is getting around the problem of delivering large genes — CEP290 (an LCA gene), for example — that won't fit in the human-engineered viruses used in gene therapy to deliver them into retinal cells.

Also, with CRISPR/Cas9, scientists are only altering the small, defective region of the gene. The rest of the gene retains all of its other natural components. By fixing only the broken part, the rest of the gene is more likely to operate as nature intended. That bodes well for its safety and efficacy.

A significant downside to CRIPSR/Cas9 is that each treatment targets a specific mutation in a gene. Many IRDs may be caused by dozens of different defects in a single gene. For example, more than 200 mutations in CEP290 can cause LCA and other related syndromic conditions. So, scientists are currently working on CRISPR/Cas9 therapies that target mutations that are relatively prevalent.

Conversely, most gene replacement therapies are designed to work regardless of how the gene is mutated; one gene therapy is likely to help most or all people with the same disease-causing gene, regardless of the mutation.

Moving into the clinic

Editas Medicine is poised to launch the first clinical trial of a CRISPR/Cas9 treatment for an IRD. Its emerging therapy targets a mutation in a region known as intron 26 of CEP290, which is a relatively prevalent cause of LCA. The company hopes to launch the human study in 2018.

Both are viable options

It is important for LCA patients to understand that the optimal treatment approach for a given patient will depend on a number of factors, including the genetic cause of the disease and available options. In some cases, gene therapy will be the best alternative — in others, gene editing may hold the most promise for saving or restoring vision.

For some patients, neither gene editing nor gene replacement will be the answer. Perhaps a stem-cell-derived treatment that works independent of their genetic profile will be best.

In the coming years, both gene-editing and gene-therapy technologies will improve and evolve, and other exciting options will emerge from IRD research labs around the world.

Visit FightBlindness.org to stay abreast of the latest research advances for LCA and other IRDs.

College Connection by Jack McCormick

Three Things Parents Can Do For Their Visually Impaired Child

Jack McCormick, left, with his Mom Kathy, Dad Bob, brother Clayton and sister Clare.

Parents want nothing but the best for their children. This is why I am so often asked questions like, "I have a 6-year-old son who has a vision impairment. What can I do to help him become independent when he grows up?" I am lucky to have the parents I do because they have done many things that have helped me become the confident and independent person I am today. I am going to share a few examples of what they did for me in the hopes that you can help someone in your life, too.

Never Say "No"
"No" was never a word I heard growing up unless I was misbehaving (not that I ever did that). Once around the age of 8, I was talking about what I wanted to be when I grow up and my Dad, a financial planner, said "Jack, maybe you can take over my business." My response was "I can't do that, you drive lots for meetings and I am not allowed to drive." Instead of agreeing with me, my parents simply replied, "Jack, you could just have the clients come to you."

Always Challenge
I was never given an easy way out because I couldn't see well. Instead I was intentionally challenged, which helped me develop many skills. In my preschool years, my parents would go for walks after dinner when it was dark. Most nights I would follow them on my tricycle. I have never been able to see in the dark. My parents didn't always know this but when they found out, they didn't stop me riding the tricycle in the dark. I believe that this helped me gain the excellent orientation and mobility skills that I have today.

Be There
Children who are different are often targeted by bullies. I was bullied frequently but I always knew that I could go to my parents for help. Being there also refers to being your child's biggest advocate. Relentlessly advocate for an accessible education for your child because without that he or she will never be independent.

I hope that you can apply these examples to your own life and that you are inspired by my parent's actions — I know that I am! As always, feel free to reach out online: jackdamccormick.wordpress.com/.

Jack McCormick is a 21-year-old honors business student at Canada's Wilfrid Laurier University in Waterloo, Ontario. Jack was diagnosed in high school with Leber congenital amaurosis due to mutations in the RPE65 gene. He is also a Sofia Sees Hope Ambassador, helping people living with LCA and IRDs.

Events

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

RARE in the Square

Global Genes
January 8–10 • San Francisco's Union Square
globalgenes.org/rareinthesquare
The 2nd Annual RARE in the Square brings together RARE disease investors, industry partners and patient community leaders.

Drug Repurposing for Rare Diseases Conference

Findacure
February 27 • London • findacure.org.uk
In conjunction with Rare Disease Day, Findacure's annual conference brings together patient groups, clinicians, researchers and life science professionals to discuss the latest developments in drug repurposing and its future role in rare disease treatment.

Rare Disease Day

National Organization for Rare Disorders (NORD)
February 28 • www.rarediseaseday.org
On Rare Disease Day, millions of patients and families around the world share their stories and promote awareness of the challenges, hopes and needs of those living with rare diseases. The 2018 global theme is Research.

Visions of Community Annual Conference

Federation for Children with Special Needs
March 10 • Boston • fcsn.org/voc
The 20th annual conference welcomes families of children with special needs from all disability types between birth through age 26.

PATIENT ADVOCACY

Successful Strategies for Rare Disease Advocates

By Danielle Chiaraluce

Lawmakers on the state level need to hear from people living with Leber congenital amaurosis (LCA) and other rare diseases to help secure funding for research, patients' needs, costs associated with living with a rare disease and accessibility on all fronts.

Your voices and those of organizations representing the rare disease community need to be heard by your state senators and representatives now to prevent elimination or reduction in funding, especially in these times of tight budgets.

This advice came during a conference workshop — Successful Strategies for Patient Organizations — at the 2017 Rare Disease & Orphan Products Breakthrough Summit in October, presented by the National Organization for Rare Disorders.

There are more than 7,000 rare diseases. When our voices are added together, they can make a difference. Start making contacts now as we approach **Rare Disease Day on Feb. 28, 2018**. Plan to combine efforts with other rare disease organizations in your state and rally for cures, regardless of the disease that you represent.

Rare disease community members represent 10 percent of legislators' constituents. Know that number before you meet. For instance, if 20,000 people live in your district, 2,000 live with rare diseases.

The legislators in your state's capital work for you. Don't be intimidated; realize the impact you can have by reaching out and/or meeting with your representative or senator. Research them in advance online, find out on which committees they serve, when and where they are in session, and contact their offices regularly.

Meeting with legislators or their staff does not have to happen at their offices. Build a relationship by inviting them to key events and meeting them at theirs. Always say hello so you stand out from the crowd. Keep calling and keep emailing.

Tell them your personal rare disease stories. Talking from the heart has impact. Even if they oppose your proposals for legislation, you've offered them a worthwhile perspective that in the end may help change their minds.

Bring notes to keep on-point. Avoid making your pitch sound too complex and don't share irrelevant information. Follow up the meeting with a thank-you card and a phone call.

Also, your best friends in the world of lawmakers are their staff members. Staffers keep track of legislators' calendars and decide when meetings take place. If you do secure a meeting with staff, don't be insulted as this often is the first step in meeting with a lawmaker in person.

Remember, legislators want to do a good job representing the people in their district. They do that by receiving pertinent information from you so they can make a difference by developing, sponsoring and enacting legislation beneficial to our rare disease community.

For more information on advocacy and making connections, visit our website at sofiaseeshope.org

Successful Strategies for Patient Organizations

1. Combine efforts with other rare disease organizations in your state.
2. Research legislators in advance online, find out on which committees they serve, when and where they are in session, and contact their offices regularly.
3. Know your numbers. Global Genes and NORD are good resources.
4. Share your personal rare disease story.
5. Take notes.
6. Get to know the staffers.
7. Invite them to your events.

Successful RPE65 Trial Patient Tami Morehouse: "There's So Much To See"

By Rosanne Smyle

Tami Morehouse made research history in the Leber congenital amaurosis world, and in the nation, when at age 44 she became the oldest person in a successful LCA-RPE65 genetic therapy trial, and the first patient to receive the retinal therapy in both eyes.

It's been a long road for Tami, now 53 and living in the Cleveland area with her husband, Mike.

With no LCA diagnosis until her 30s, Tami made her way through life doing whatever she had to do. Sink or swim, she developed good coping and resource skills.

"I did what I had to do," she said. "I had enough vision to make it work."

No one noticed her vision problems until she was about a year old, when nighttime came, and she couldn't see well at all. Attention focused on the idea she had night blindness.

It is difficult to detect vision clarity in small children, but her mom and dad knew something wasn't quite right. Doctors picked up on it when she was 5, during an exam in which she recalled she was scared to death, screaming and crying, because they were poking around her eyes.

They determined she had issues with the clarity and clearness of vision, known as visual acuity.

From kindergarten into her 20s, she adapted to her surroundings and to her level of vision. She had difficulty seeing the chalkboard, reading a paper or deciphering bulletin-board postings.

"All of my teachers knew I had a hard time. I needed more light than the average kid. I remember the hallways of my elementary school were very, very dim and I had a hard time making my way around."

Something her father said a long time ago helped her along the way. Her dad, who always called her Timmer, said, "Timmer, we all have our troubles, and if you want me to take you anywhere or do anything, just ask me.' They just treated me like the typical kid I was not."

Studying social work at Edinboro University in Pennsylvania, Tami scheduled classes in familiar and comfortable places. She avoided night classes, and when she couldn't, she'd figure out how to get there, walking in better lit areas.

She realized that all her life she was a tweener — someone poised in-between.

"I was a tweener for forever because I did most things like everyone else did and there was that part that everyone else didn't know."

Since she was young, some sort of eye doctor tracked her through the years, but it wasn't until she got her first social-work job and her then-boyfriend, now-husband, Mike, said they needed to explore potential options.

Specialists diagnosed her under the retinitis-pigmentosa umbrella and told her she would lose her vision. They said it was good she already had her education and she should consider having her children now. "It was a heartbreaker day," Tami recalled. As she aged, so did her retina and its ability to function well. She went from reading storybooks to her three children to reading Dr. Seuss alphabet books. Some days she saw only hazy grays and browns and needed the brightest lights to see.

"It was really very scary. At that point, before the trial, I had more poor-vision days, than OK-vision days. Sometimes I was scared to death to set my alarm. What if tomorrow is the day I wake up and my vision doesn't get better? What if I wake up and I can't see?"

But one Sunday morning, Tami was in the house, and her husband was out in the garage. He suddenly barreled in. He had heard on the radio that a study was being done on children with LCA in Philadelphia by Dr. Jean Bennett and Dr. Albert McGuire. Mike called Bennett the next day, and the wheels turned fast. Bennett opened the study at Children's Hospital of Philadelphia to older patients and invited Tami to take part in the trial that would change her life.

In March 2009, doctors injected her left eye with healthy cells to help her retina perform more efficiently and regenerate healthy cells.

Several days later, she and her husband were having dinner in Philadelphia and Tami reached over and picked up her drink.

'Do you know that you just reached over and picked that up, you didn't feel for it?,' Mike asked. 'You just looked out and saw it and picked it up!'

The injection in her right eye in November 2010 brought more brightness and clarity, to the point where she could see some eye-chart letters.

In spring at a baseball practice for one of her sons, she noticed colors more than ever before.

"It was color and light and movement and the kind of stuff people take for granted every day, which may seem small if you have it. Once you lose those little things and then get them back, you realize how important they were. For me it was huge."

Tami also got to see her daughter taunting the opposing pitcher on her softball team, as she frequently danced up and down the third-base line to almost always steal home.

Her only wish was that the therapy had been an option sooner, because as the years passed, her retinas kept deteriorating.

"If this was a life-threatening illness," she quipped, "I would have died a long time ago."

She advises anyone with LCA-RPE65 to investigate whether the therapy is an option, saying, "Time is of the essence. Don't give up. There were a lot of us in that trial and we all seem to have different levels of benefits from procedures, whether you have a little vision or a lot of vision. I just value my vision so much, I just think everybody needs to act and respect whatever level you have and just be glad to have it. There's so much to see. It's an incredible gift."

You can read more about Tami's story on our website, sofiaseeshope.org.

Sofia Sees Hope Lauds FDA Approval of LUXTURNA™

LUXTURNA™ (voretigene neparvovec), is the first pharmacologic treatment for inherited retinal disease and the first gene therapy for a genetic disease in the US

On Dec. 19, 2017, the U.S. Food and Drug Administration approved LUXTURNA™, the first gene therapy for a genetic disease in the United States.

Spark Therapeutics' LUXTURNA™ (voretigene neparvovec) is a potential one-time gene therapy for the treatment of patients with vision loss due to confirmed biallelic RPE65-mediated inherited retinal dystrophies. The RPE65 gene mutation is within the Leber congenital amaurosis (LCA) family of inherited gene mutations that eventually lead to total blindness.

LUXTURNA™ is the first FDA-approved gene therapy for a genetic disease, the first and only pharmacologic treatment for an inherited retinal disease (IRD) and the first adeno-associated virus (AAV) vector gene therapy approved in the U.S.

"This is a huge day for RPE65 patients, and indeed for everyone in the LCA and IRD communities," SSH co-founder Laura Manfre said. "While this first treatment does not address the dozens of other genetic mutations that cause blindness for LCA and IRD individuals, we can hope that this will in turn pave the way for continued research and treatment for our entire LCA and rare IRD community. This marks another positive step forward on the path to changing the lives for many people."

Manfre added that Sofia Sees Hope will expand its outreach and support programs in the new year to provide information for patients and their families about what happens now for the RPE65 population as well as other genetic mutations and the large still-undiagnosed population. Plans currently include access to free genetic testing programs, supporting family connections and continued education and awareness.

"As an advocacy and education organization, we will be providing information on our website and social media channels," Manfre said. "We are also pleased with the patient support Spark is providing to the RPE65 community."

Read more about LUXTURNA's approval and what it means to the IRD community at www.sofiaseeshope.org

The *Seeing Hope* newsletter
is published quarterly by Sofia Sees Hope, 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.

To learn more about Sofia Sees Hope visit our website at www.sofiaseeshope.org.

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