USHER 1F COLLABORATIVE

Newsletter

Fall - Winter 2018

HOW FAR WE HAVE COME AND HOW FAR WE HAVE TO GO

When we began Usher 1F Collaborative in December 2013, we had $2000 in the bank and one researcher who had Usher 1F as a future initiative. Our initial goal was the development of an animal model on which to test potential new treatments. Five years later, we now have two animal models a zebrafish and a mouse, both of which share all three human Usher 1F traits, deafness, poor balance, and progressive vision loss, and six research groups actively working on a cure at academic medical centers that include the University of Oregon Institute of Neuroscience, the University of Maryland, the University of Iowa, Harvard University, the University of Western Australia, and the Centre for Eye Research Australia, with a research group at the University of Pittsburgh eager to join us as well. We have identified three potential pathways to a cure, drug treatment, gene therapy, and stem cell transplantation, and are funding research in all three areas. Our first gene replacement therapy is nearly ready to be tested in our mouse model.

We reached another milestone this year with the awarding of our 2018 grants in May, surpassing the $1 million mark in Usher 1F Collaborative provided research funding. The good news continued as the team at the Centre for Eye Research Australia obtained a substantial grant from the Australian government specifically for Usher 1F vision research, and a Harvard researcher, whom you will read about in this newsletter, obtained substantial funding to develop gene therapy for Usher 1F. All of these initiatives combined now equal over $3 million in research to cure the vision loss of Usher 1F.

How You Can Help

We have made great progress, but our work is not yet done and will not be done until every person with Usher 1F can see and can continue to see for a lifetime. Our investigators are working on multiple paths to a cure, but their pace and scope of work are limited by our ability to fund their programs and recruit new investigative teams. Our most immediate needs are, first, $200,000 to have our University of Oregon team perform a drug throughput screen to test multiple drugs, including some that are already FDA approved for other uses. If one or more are identified as effective on our zebrafish, then testing on humans, especially for those drugs that are already FDA approved, would be the fastest path to a treatment. Second, we want to bring on board a new researcher from the University of Pittsburgh who has developed a unique and effective way of delivering our large gene. She has submitted a proposal that will cost $100,000 to fund. This total of $300,000 is above and beyond the work that we are already funding and that we need to continue to fund in order to maintain our progress.

With your support, we can maintain and grow our momentum so that our team will be closer to ensuring that no child born with this disease will face a future of blindness, and no adult will live in darkness. We are eager to see what the next five years will bring and invite you to be part of the cure.

FINLEY

By Natasha Mercer

Finley is a little boy with Usher 1F who lives with his parents in Newfoundland, Canada

Don't make the house too quiet when he naps, or you will get him used to sleeping in complete silence, then he will never sleep. This was a different type of parenting advice than the typical "sleep when the baby sleeps." It was the type of parenting advice that made sense, the kind that came from experienced parents, the ones that truly knew what they were talking about. We expected blonde hair, blue eyes, lots of nursery rhymes and even more books. When Finley was in utero, we vowed we would read books to him constantly. We were dedicated to giving him the best opportunity to grow and develop into the smartest little boy - a boy who wanted to learn and thrive for nothing short of excellence.

When we got the news that Finley was deaf, my mother's intuition kicked into overdrive. I had a gut feeling that there was more to it. Months later, I was proven right and now here we are. Finley's dad was speechless, motionless, sick to his stomach, and had a mixture of feelings, like guilt. He questioned why him? Why Finley? What's next? What have we ever done to deserve this? We all want what is best for our children. We want happy, healthy and normal. We didn't expect the countless hours of driving to and from appointments. We didn't expect 60,000 KM a year. We didn't expect the 25 physicians/ specialists/ therapists. We both work busy schedules. Our days off and vacation days are spent at doctors' offices, but now more than ever we are dedicated to helping Finley take advantage of every resource that we can get our hands on.

There was no family history of genetic mutations, no history of deafness, blindness, nothing. No one expected this journey. But that is common with Usher Syndrome - you don't expect it until it is you, your baby, your grandchild, your best friend. We had no idea what Usher Syndrome was, and now? It is everything we think about, every time we start our car engine, every time Finley looks into our eyes, every time we see babies younger than him walking, when our angel can't yet stand, we mourn our baby's future every day; often the sadness consumes us, but Finley is a 16-month-old who is the sweetest, most generous, pleasant child we have ever laid eyes on. His smile would light up a room. He graciously cuddles with everyone. He always shares, even his most favorite snacks; he loves to paint, drive his quad, bake cookies with mommy and daddy, and he loves everything that has wheels. With his cochlear implants, he loves to hear us sing, but more than anything, he thinks it is the funniest thing ever when the smoke detector goes off when daddy is cooking supper.

A lot of people stare at us, and some will even point when we are out in public, and with his cochlear implant retention devices he is often mistaken for a girl. We always explain that his cochlear implants help him hear like glasses help people see. But truthfully, it is glasses that allow people to see with poor sight, but not blindness - just like hearing aids help people with some hearing loss hear, but not deafness. We have cochlear implants for deafness, so now we need a cure for blindness, for our children, for our family, both present, and future.

NEW USHER 1F RESEARCHER, DAVID COREY, PHD

David Corey, PhD., is the Bertarelli Professor of Translational Medical Science, Department of Neurobiology, Harvard Medical School. He has previously studied the Usher 1F gene and has now taken on developing gene therapy for the vision loss of Usher 1F. Below, he describes this new project.

A major gift from the Bertarelli Foundation to Harvard Medical School will support four new grants on understanding and treating sensory disorders. One of these grants, for work by scientists in Boston and Switzerland, will fund research into new strategies to treat the loss of vision in Usher 1F. Principal investigators of the team are Botond Roska, an expert on the retina who is located at the Institute for Ophthalmology in Basel, Switzerland, and David Corey and Artur Indzhykulian, of Harvard Medical School and Massachusetts Eye and Ear, who have studied the structure and function of protocadherin-15 in the auditory system and who have pioneered new methods for gene therapy for other forms of hereditary deafness.

Traditional approaches for gene therapy for recessive disorders often involve using AAV viral vectors to deliver DNA to affected cells; the DNA allows the cells to make normal protein, which may compensate for the defective inherited form. AAV is a small vector, however, which holds only about 4,700 individual bases of DNA. Protocadherin-15 requires at least 6,000 bases—too long to fit in AAV. The Roska, Corey and Indzhykulian laboratories will collaborate to try to develop new ways to overcome this limit, and they hope eventually to halt the loss of vision in Usher 1F patients.

OVERCOMING THE CHALLENGES OF LIFE WITH USHER 1F

By Ryan Bandroff

Ryan Bondroff is an adult with Usher 1F. He describes a few insights into his life as a deafblind person.

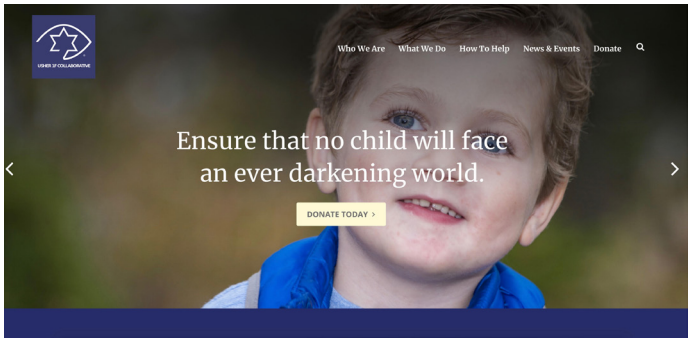
Usher Syndrome 1F does not stop me from accomplishing so many things. I have a national professional job with a national telecommunications company where I am a consumer relations manager for the DeafBlind community. With supports, I travel extensively for work, and I love to travel and meet new people. On a recent vacation, I traveled to the United Kingdom with my DeafBlind friend. We traveled without having a Support Service Provider (SSP), which was fun and challenging. I am active in the DeafBlind community in various ways.

Without a cure, I need the SSP program that is designed to provide communication access/visual/environmnental supports to the Deafblind people who need it the most. It is a very, very long way to get funding from the government to support the SSP program. We rely heavily on others to support our needs, which is not a good thing to do.

SECOND USHER 1F RESEARCHERS MEETING PLANNED

In May 2019, Usher 1F Collaborative will host our second Usher 1F researchers meeting in conjunction with the annual meeting of The Association for Research in Vision and Ophthalmology (ARVO) in Vancouver, Canada. Following the success of our previous meeting in May 2017, which provided our researchers with the opportunity to share their work and ideas, they had requested a chance to repeat the experience to enable them to share updates and progress. So far, planned attendees include team members from the University of Oregon, the University of Maryland, Harvard University, and the Center for Eye Research Australia. We look forward to the progress that will result when we put all of the great minds working on a cure together to discuss and share strategies and will report on the outcome in our June 2019 newsletter.

CHECK OUT OUR NEW WEBSITE!



Check out our bold, beautiful, newly redesigned website at **https://www.usher1f.org**, where you can learn about our work to find a cure and about those who are affected by Usher 1F.

Photo caption:
Clockwise from top left: Tarzana, CA; San Francisco, CA; Chestnut Hill, MA; Deerfield, IL; Wyckoff, NJ; Atlanta, GA

SIGHT.SOUND.CYCLE 2018

Sight.Sound.Cycle was, once again, a great success. Participants cycled in seven locations around the U.S., raising $100,000 for Usher 1F research. A great big thank you from all of us to all who participated, particularly to those who hosted events.

SEND DONATIONS

Usher 1F Collaborative
#228
321 Walnut Street
Newtonville, MA 02460-1927

www.Usher1F.org/Donate