USHER 1F COLLABORATIVE

Newsletter

Spring 2018

PROGRESS TOWARD OUR DREAM OF A CURE

> Every great dream begins with a dreamer. Always remember, you have within you the strength, the patience, and the passion to reach for the stars to change the world.
>
> ~Harriet Tubman

We have succeeded, in the four years since we began, in substantially escalating the scope and pace of research for a cure for the vision loss of Usher 1F. While in 2013, only one research lab had Usher 1F in its game plan, we now fund significant research at four research labs, with research at two other labs also advancing in part because of our efforts. Most recently, with the awarding of our 2018 researchers' grants, we reached the milestone of over $1 million funded since our inception. Over the past year, this funding has allowed our researchers to begin making substantial inroads toward a cure. Notable accomplishments over the past year include:

• Development of two knock-in animal models, a mouse and a zebrafish. A knock-in model is one that has the actual human genetic mutation inserted vs. having a similar animal mutation. What is significant about both of these animal models is that, for the first time, we have Usher 1F models that display not only deafness and vestibular issues but also progressive vision loss. Thus, these models have provided us with a means of testing and measuring the effects of potential treatments to rescue vision.

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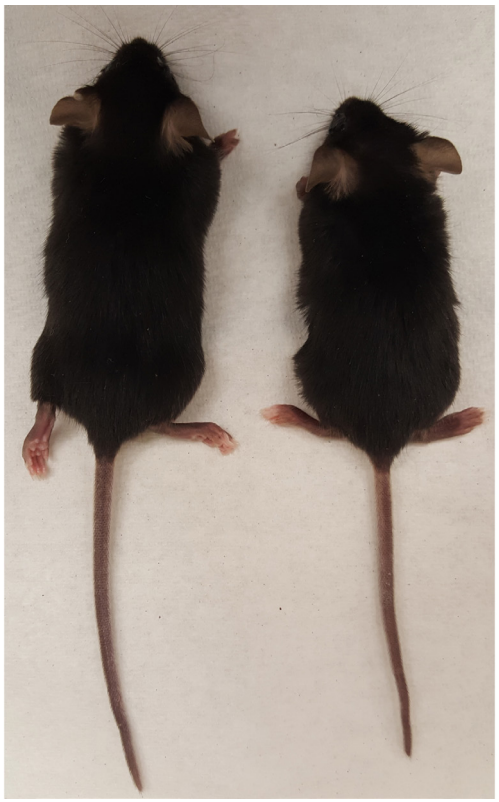


• Testing begun of potential drug treatments on our fish and mouse models. We are very excited to report that one of our researchers has realized success in preliminary testing of a drug and will be conducting further testing this year to verify these results.

• Progress toward development of two types of gene replacement, a split gene and an abbreviated gene, similar to the recently FDA approved Luxturna for another inherited retinal disorder.

• Development of a viral vector that has the potential to deliver our entire large gene.

• Preparation to complete an FDA application to begin a clinical trial using iPSC derived photoreceptor precursor cells for a similar inherited retinal disease (iPSCs are induced pluripotent stem cells, i.e. those derived from patients' own skin cells).

• A much needed natural history study in progress, which will document the natural course of vision loss in Usher 1F. In order to show efficacy of a treatment, this is a critical step.

Our donors have been instrumental in helping us achieve these milestones. We are so grateful for your support. While we have achieved so much, getting a successful treatment from the first stages of development and testing in the lab to and through a clinical trial to FDA approval is a lengthy process. Some aspects of research cannot be rushed. Here is where you can help.

• New equipment that can speed the pace of research is expensive, often beyond the reach of existing lab funding.

• Some tasks that could be performed simultaneously have to, instead, be performed sequentially because of insufficient dedicated lab personnel.

While we cannot rush animal development, including time to gestate, grow, and reproduce, funding to support new equipment and additional lab personnel can accelerate the pace of Usher 1F research.

We continue to develop more than one path to a cure so that all with Usher 1F, regardless of stage of vision loss, can realize their dream of continuing to see our world. Thank you so much for your support. We look forward to having you continue with us on our journey toward our dream of a cure!

ANDI, AARON, CHASE, AND JUANITO NEED A CURE!

Meet four of our youngest with Usher 1F, Andi, Aaron, Chase, and Juanito, whose parents are fervently hoping for a cure.

Juanito

By Gabby Meza, Juanito's mom

Juanito is a jolly 19-month-old and the youngest of our blended family of seven children. His lack of vestibular ability does not stop him from climbing everything! He's an inquisitive little boy who loves to interact with everyone, and even though he doesn't walk just yet, he stops at nothing to get around everywhere with his little walker. Juanito was diagnosed at just a couple of weeks old with bilateral profound hearing loss. Receiving such a diagnosis was, to say the least, shocking, as we were not aware of any hearing loss within the family. But my husband and I clearly remember thinking, "No big deal, we've got this." Right before Juanito turned a year old, we were able to confirm through genetic testing that he had the rare genetic disorder Usher Syndrome type 1F. That news was not received lightly. For the next days

we went through a period of mourning and just flat-out disbelief. Thankfully, my husband and I are on the same page with moving on from lamenting and are solely focused with providing him all the love, confidence and enthusiasm he needs to adapt to his life with cochlear implants and future vision loss. His success is incumbent upon us, so we try to maximize the utilization of resources currently available for him through various agencies. As he continues to grow older, we will reassure him that great work is being done to help people with many different sight conditions, including his own. It is vital to us as his parents to stay informed of new research, different support groups, and new advancements in the fight against sight loss, as this will help him overcome the challenges he will be facing every day and will give him hope for the future.

Andi and Aaron

By Jessica Picanzo, Andi and Aaron's mom

This is Andi and Aaron Picanzo. Andi is a beautiful, outgoing two-year-old. She loves to play outside, sing and dance to music, and help take care of her little brother! When Andi failed the newborn hearing screening, we followed through with genetic testing to find out the cause of her hearing loss. We learned when she was eight-months-old that Andi has Usher Syndrome type 1F. Two months later, she had cochlear implant surgery and has far exceeded our expectations!

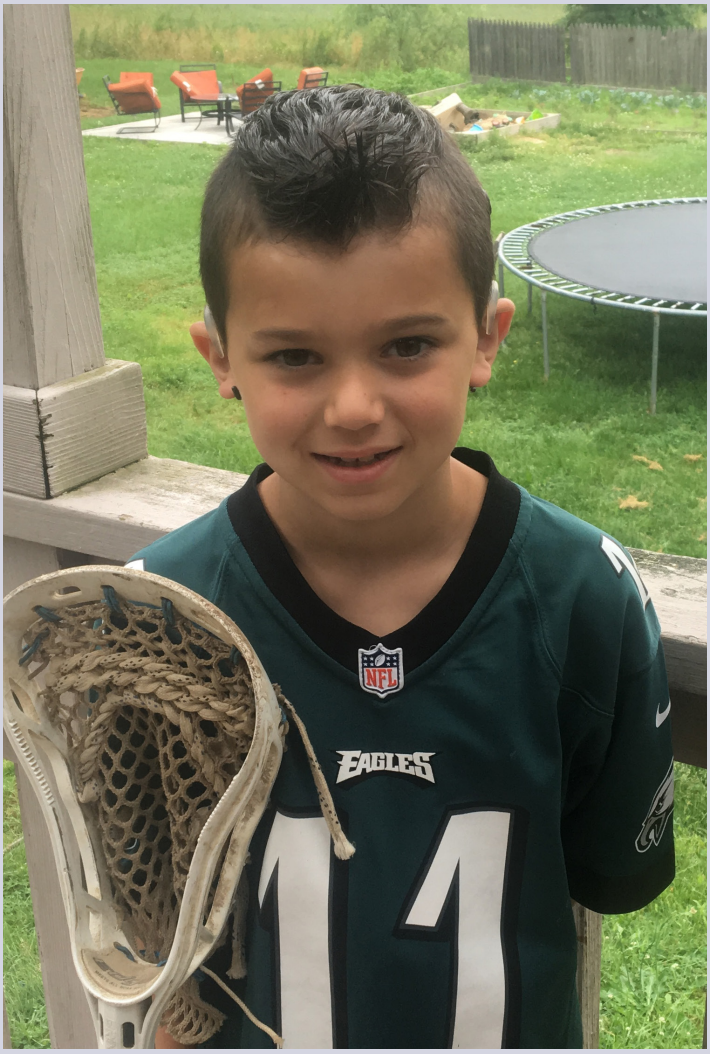
Aaron is two-months-old and also has Usher Syndrome. Finding a cure for Usher Syndrome would mean the world to our family and give Andi and Aaron the chance at a normal childhood and life without having to wonder when their world will go dark. We are desperate but hopeful for a cure for our children.

Chase

By Ally Doerrmann, Chase's mom

Meet Chase! - An amazing 7-year-old who was diagnosed with Usher Syndrome type 1F in July 2017. Chase is in first grade, plays baseball, basketball, soccer, and each year the list gets longer (one tired mom).

I knew in my heart when I noticed Chase having trouble in the dark on a family camping trip to Knoebels that something was wrong. Now, two years later, Chase is still rocking it and an all-around great kid who loves sports, swimming, dirt bikes, hunting, fishing, and being with his friends. As devastating as it was for my husband and me to find out that Chase has a disease that could eventually take his vision, and our hearts broke to think he might not be able to do the things he accomplishes now, we know anything is possible and we will stop at nothing! The Ushers' community has been nothing short of amazing. The day I found out about Chase, I had other moms message me right away. I never felt alone and immediately found comfort in them. We can only hope for some sort of treatment or cure because no child or adult deserves to lose their vision, but that will not stop us from trucking through life with smiles on our faces.



SIGHT.SOUND.CYCLE 2018 IS COMING IN SEPTEMBER!

On September 29th and 30th, for the third year, we will be spinning for a great cause, raising money and awareness for Usher Syndrome Type 1F. We MUST find a cure, so come and spin.

Expert, beginner, wannabe spinner – It doesn't matter your level. All proceeds go directly for research through Usher 1F Collaborative, a 501(c)3 nonprofit foundation.

DON'T MISS YOUR CHANCE TO HELP!

For more Information, visit http://usher1f.org/sight-sound-cycle/

Sign up beginning in July.

SEND DONATIONS

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www.Usher1F.org/Donate