USHER 1F COLLABORATIVE

NEWSLETTER
Spring/Summer 2025

Matti

USHER 1F IN GERMANY:

Meet Matti

Hello, I'm Sabrina (43). I live with my children Neele (17), Lotta (15), and Matti (3) in Gäufelden, a small town in Baden-Württemberg, Germany. Matti is my third child and will turn 4 at the end of May. He's a funny little guy with tons of energy—which sometimes pushes his older sisters to their limits.

I've been mostly on my own with the bunch for about a year-and-a-half now, since their dad and I separated. Did our very exhausting and emotional experiences contribute to that? No one really knows.

Matti was born on May 30, 2021, at the Herrenberg hospital, south of Stuttgart. His start in life was a bit rough. It was a difficult birth that ended in an emergency C-section, so his dad had to take care of him first until I was able to again. When the routine hearing screening was done, it repeatedly showed no results. But the nurses and doctors told us not to worry. Was it broken equipment, incorrect handling, or just a restless baby? No one could say.

We were discharged and advised to see a pediatric ENT, but they also couldn't give us any answers. It didn't feel right, so we looked for a specialist who fairly quickly diagnosed Matti with severe hearing loss. That was the first moment it felt like the ground had been ripped out from under our feet.

At six weeks old, Matti got his first hearing aids. But they never really fit properly, constantly whistled, and became a huge burden, especially since he didn't really accept them. As we would later learn, it was because he wasn't benefiting from them.

The audiologists always saw great reactions during the constant adjustments of the hearing aids. At first, we had to drive about an hour every week with the little one for appointments. But at home, we noticed no reactions. Nothing bothered him—not the doorbell, not the vacuum cleaner, not a door slamming shut due to a draft.

Fortunately, we were referred to the Olgahospital in Stuttgart. More tests were ordered. A hearing test under general anesthesia, called a BERA, brought a sobering result. Matti's auditory nerve didn't respond. The measurements stopped at 100 dB. Our child was deaf.

Pretty quickly, we began talking about the possibility of cochlear implants. I still hoped it was all just a big mistake.

So, the first surgery was at 11 months, the second shortly after his first birthday. Everything went well. The wounds healed nicely. He accepted the devices, and we thought we were on a good path.

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We were told we could do a genetic test to find out why Matti is deaf, if we wanted to. We quickly decided that we did want to know the cause—with all the consequences. Or so we thought.

The genetic test was ordered, and just before Christmas 2022, we were invited to the clinic for a discussion. We went to the clinic totally naïve, thinking we'd just get the result and then head to the Christmas market with the whole family. It was practically around the corner.

WE REMAIN POSITIVE. MATTI IS THE SUNSHINE OF OUR FAMILY.

But in the hallway, I already knew something wasn't right. We could see into the geneticist's office as he read our file. His body language didn't look promising—and I was right.

Inside, they explained that they had found a genetic defect, passed down 50% from me and 50% from his dad. Matti has Usher syndrome.

Now the condition had a name. But what did that mean for us? Matti will go blind—likely in his childhood. He has one of the most aggressive forms of Usher syndrome: Type 1F.

Our world collapsed. It was the worst day of my life. You go numb from pain and uncertainty. Your heart just breaks for this little guy.

No one can imagine what it's like unless they've been in this situation themselves. Your child's future is essentially written, and you have no say in it. "Helpless" doesn't even come close to describing it. You feel punished. You were handling the deafness well—why this too?

Strangely enough, I had seen something about Usher syndrome years ago and was horrified by it. How can it be? Deaf and blind? What a terrible combination.

Will my child be able to lead an independent life? When will the vision problems start? Will he need to go to a special needs school?

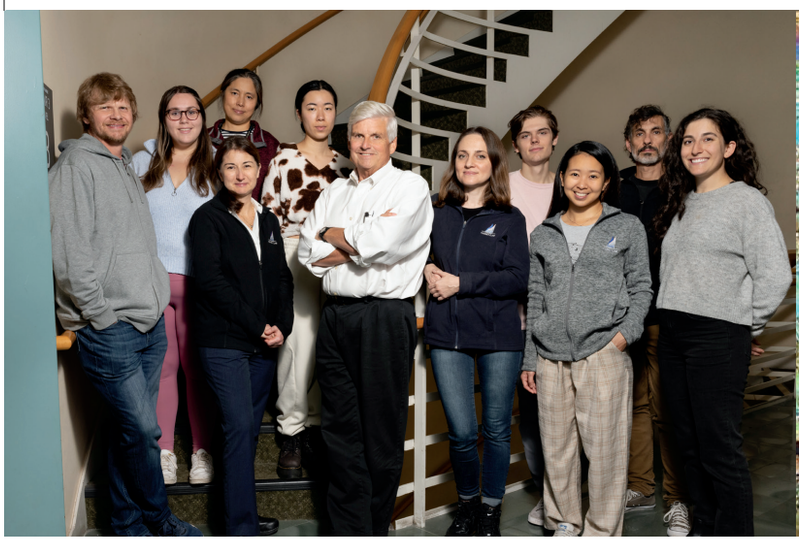
All we can do is prepare our little one as best we can. Lots of normalcy, outings, fun, toddler gymnastics, goofing around—combined with the necessities of our new daily routine: physical therapy, occupational therapy, speech therapy, eye doctor appointments, hearing aid adjustments at the hospital, rehab stays, and more.

Matti has shown me that life is absolutely worth living, even with limitations. Right now, we're lucky that he has his balance issues well under control. He goes to a forest kindergarten with 20 kids. His needs are lovingly addressed. Our next step is school enrollment—hopefully still without vision impairment.

Because of Matti, I've become active in fundraising for research on Usher syndrome. It's a way for me to cope with the diagnosis. You don't feel helpless anymore. You're doing something.

And the hope for a treatment for Usher syndrome is strong. ◆

Matti and Sabrina with their family



Members of the Corey Lab

Members of the Westerfield Lab

Spring 2025 Research Update

In 2022, the Usher 1F Collaborative Board of Trustees voted to focus our funding on projects that have a clear path to the clinic, that is on translational rather than basic science research. Where initially, we had cast a wide net, we have now narrowed the number of our funded research labs to those that meet this goal.

Harvard Medical School, David Corey Lab:

Preclinical testing thus far of our Usher 1F mini-gene has yielded very promising results. Currently, the team is analyzing data and preparing papers for publication. We will announce these papers when they are published and will summarize the results.

Usher 1F Collaborative has funded a research technician, Annie Chen, to work full-time on the Corey Lab's Usher 1F research. Annie's work has been invaluable in speeding the pace of lab's Usher 1F research. She will be leaving at the end of the summer to begin a PhD in Neuroscience at Harvard. While this is a loss for us, we congratulate Annie. Because of the current situation with government funding for Harvard University, which includes the medical school, Harvard has instituted a hiring freeze. Thus, we will need to fully fund the cost of a replacement technician for Annie or face slowing our progress.

Harvard Medical School, David Corey Lab, and The Salk Institute for Biological Studies, Samuel Pfaff Lab:

As reported in our Fall 2024 newsletter, Usher 1F Collaborative has funded a promising research collaboration between the Corey Lab at Harvard and the Pfaff Lab at The Salk Institute to use a revolutionary approach to delivering our large gene to the retina. We are pleased to report that the Corey Lab now has the vectors from the Pfaff Lab and has begun testing in their mouse model. David Corey says of this partnership, "We are very excited to be testing Sam Pfaff's new method for delivering large proteins like PCDH15, and I would never have known about it if the Collaborative had not brought us together."

The University of Oregon Institute of Neuroscience, Monte Westerfield Lab:

"With funding from the Usher 1F Collaborative, we have generated several zebrafish models of human USH1F mutations. These zebrafish models recapitulate the symptoms of USH1F human patients accurately, including deafness, balance problems, and vision loss due to retinal degeneration. Zebrafish develop very rapidly, and USH1F symptoms appear in mutants within the first week of life. Moreover, mutant adults can provide hundreds of animals that we can study in each experiment. These features have allowed us to design a drug screen for compounds that can be used in therapies to treat vision, hearing, and balance problems in USH1F. We have one promising candidate and are testing additional potential therapeutics." ◆

~Monte Westerfield

JORDAN MELIKER'S

Gift of Hope

In 2024, family and friends of Jordan Meliker gathered to celebrate a milestone in his life as he became a Bar Mitzvah. But Jordan's preparation extended far beyond the bimah. As part of his Bar Mitzvah, he took on a mitzvah project – an opportunity to give back and make a difference.

Inspired by his dear family friend, Zachary Root, Jordan organized a fundraising event to raise money and awareness for Usher 1F Collaborative with the hope that medical research will change the future of his friend and save Zachary's vision. Guests at the event wore glasses that simulate blindness while they attempted to paint on canvas.

Jordan wrote, "Zachary has overcome many challenges related to Usher 1F, but one that he continues to fight is the challenge to maintain his vision. Zachary's vision is slowly deteriorating and ultimately, he will lose his vision unless a cure is found."

Jordan's friends and family responded, with 55 supporters raising $3,081 for Usher 1F Collaborative. ◆

Jordan Meliker and friends at his Usher 1F fundraising event

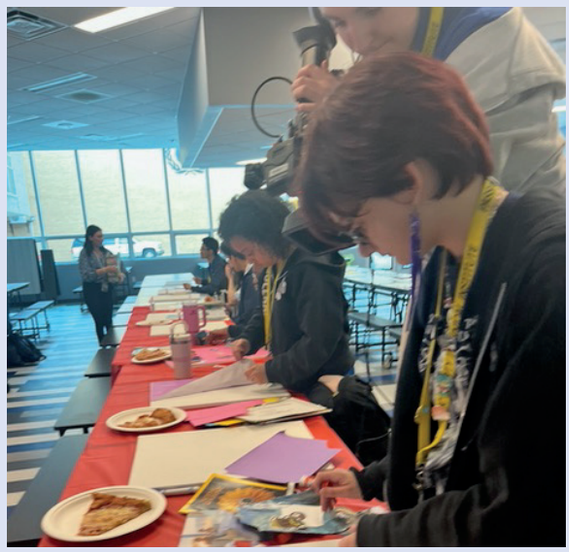
Jordan Meliker and friends

EYES ON THE FUTURE:

Students Create Vision Boards for Usher 1F

In 2024, students and teachers at Passaic County Technical-Vocational Schools (PCTVS) in Wayne, NJ, held a fundraiser for Usher 1F Collaborative. After raising money, students gathered and wore blindfolds with a small pinhole to simulate vision loss from Usher 1F and painted artwork. PCTVS students are back at it this year, and they wanted to once again raise money for Usher 1F Collaborative.

This year, students are creating *Vision* Boards, setting goals for their own bright future, after they help raise funds to create a brighter future for those living with Usher 1F. The students raised more than $1,200 and then gathered to create their Vision Boards. ◆



PCTVS students creating their vision boards

PCTVS students sharing their completed vision boards

Gary and Laurie Shapiro

Honoring a Legacy of Love and Support for Usher 1F

Last year on Giving Tuesday, board members Margi Levitt and Dorie Shapiro planned a special evening to honor the late Laurie and Gary Shapiro in support of Usher 1F Collaborative. Margi and her husband Tom opened their Paradise Valley, Arizona, home to 80 guests, who remembered the Shapiros' legacy of tirelessly supporting Usher 1F Collaborative and its goal to treat the disease. With the Levitts, twelve other couples co-hosted the event, including Dorie's brother and sister-in-law, David and Elizabeth Shapiro. Attendees enjoyed wine and hors d'oeuvres, a silent auction, and learned more about the mission of the Collaborative through the short film, *Seeing Forward*. At the Giving Tuesday event, guests were moved to contribute $42,000 to the Laurie and Gary Shapiro Memorial Fund, which was established earlier in the year. In all, donors contributed $114,000 to the fund in 2024, which supports the ***Seeing Forward*** initiative. The Shapiros' legacy endures through the generosity of a committed community. ◆

Dorie Shapiro with her brother David and sister-in-law Elizabeth

Forte Family at Catch the Ace with Doug Anthony (right) of the Kin Club of Russell

KIN CLUB OF RUSSELL:

Catch the Ace lottery

By Nicolas Forte, Usher 1F Collaborative Canada

I first learned of the Catch the Ace (CTA) Lottery with the Kin Club of Russell on our local news. They were reporting on the possibility of winning a jackpot of $890,000. That, coupled with interest from my colleagues, sparked an idea in me – What if Usher 1F Collaborative Canada was included in this progressive lottery fundraiser?

The goal of this progressive lottery for Ontario residents is to raise funds for Ottawa area charities while giving people an opportunity to win a large sum of money. I got straight to work connecting with the CTA team, submitting our application, and they accepted!

With the help of social media and effort from family, friends, and colleagues, we've exceeded 1000 tickets sold! The race is still on - if you live in Ontario, grab your ticket today to Catch the Ace.

Check @usher1fcanada on Instagram and Facebook for updates. We're grateful to be a charity chosen, and with your support and a touch of luck, we're making things happen across the country to find a cure. Join us in this fight! ◆

USHER 1F COLLABORATIVE

SEEING FORWARD

OUR 3-YEAR MAJOR-GIFT FUNDRAISING INITIATIVE TO MEET OUR NEXT BOLD GOALS ON A RAPID PATH TOWARD A CURE

FUNDRAISING UPDATE

$0 **$1,012,657** $3,015,000

updated on May 7, 2025

usher1f.org/seeingforward

ILLUMINATING THE FUTURE:

Canada Invests in Sight and Science

We are excited to launch a campaign offering Canadians the unique opportunity to lead the scientific research for a drug repurposing study that could have a tremendous impact on the lives of those with Usher 1F.

WHAT IS DRUG REPURPOSING?

Identifying new purposes for existing drugs with a proven safety record is the fastest path to the clinic for Usher 1F patients. An existing drug may slow the progression of their vision loss, but we must do a drug screen to identify which drug(s) will work. We will issue a Request for Proposals to Canadian research scientists to execute this study.

WHY NOW?

The time is right for Canadians to lead the effort. The National Strategy for Drugs for Rare Diseases was adopted by every province in April 2025. With all provinces and territories recognizing the need for treatments for rare diseases and committed to investing in innovation, the time is right now!

FOR MORE INFORMATION ON ILLUMINATING THE FUTURE: CANADA INVESTS IN SIGHT AND SCIENCE, PLEASE VISIT **USHER1F.CA**.

USHER 1F COLLABORATIVE

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