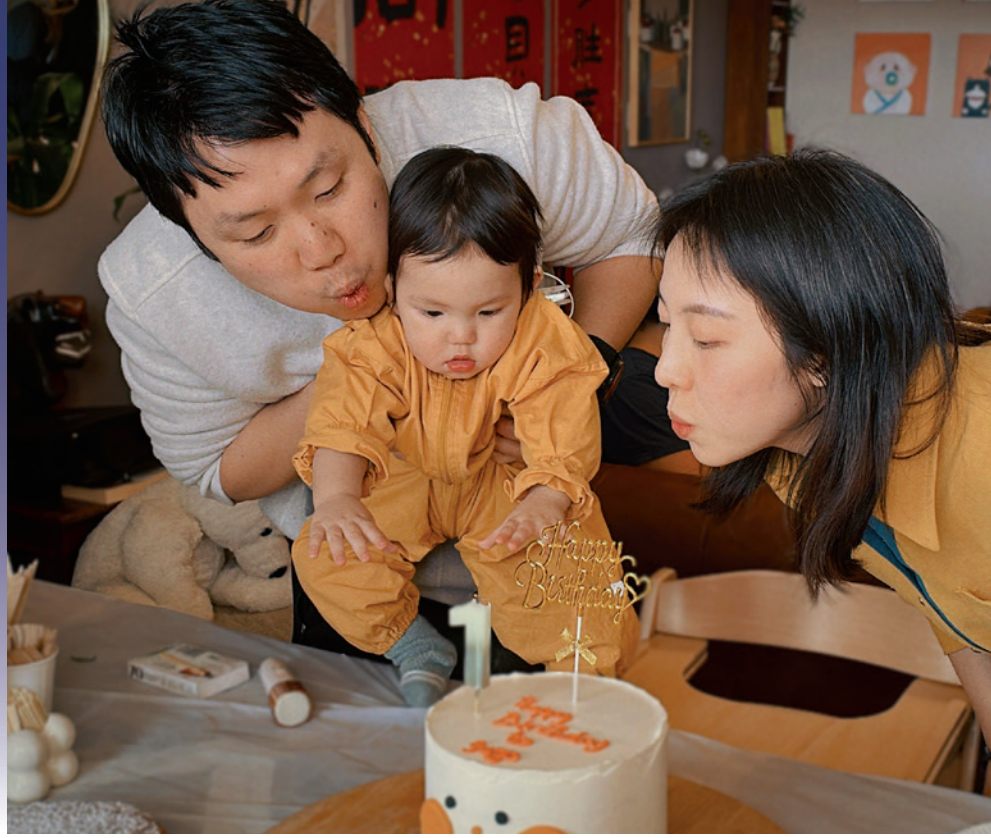
USHER 1F COLLABORATIVE

NEWSLETTER
Fall 2025



Yuge with his parents Rongkun and Xin on his first birthday

YUGE'S STORY:

Courage, Love, and Hope with Usher 1F

Our baby, Yuge Wang, was born on January 21, 2024, in Geneva, Switzerland. According to the Chinese zodiac, he is a rabbit. Sadly, this little rabbit was born without sharp ears—he did not pass the newborn hearing test at the hospital.

Because there was no history of deafness in either of our families, we clung to the hope that the test must have been inaccurate. But once we were home, we did notice that he didn't respond to voices or to the sound of objects dropping. Our anxiety grew. At his follow-up test two months later, doctors at Geneva University Hospital confirmed what we feared: Yuge was born with profound hearing loss. Listening to the doctor's diagnosis, I felt my heart shatter. Tears streamed down my face as I wondered how my child would face a world without sound.

Reality didn't allow us to stay in grief for long. The team of specialists at Geneva University Hospital immediately stepped in, reminding me again and again that an early diagnosis and intervention meant Yuge would still have the chance to hear and speak like other children. They quickly prepared a cochlear implant plan and arranged balance tests, speech therapy, genetic testing, and vision screening. (We later learned that they had treated another child with Usher syndrome type 1B four years earlier, and based on Yuge's symptoms, they already suspected a similar diagnosis.)

On November 11, 2024, the genetic test confirmed it: Yuge has Usher syndrome type 1F. This meant that on top of his deafness, our little boy would also face progressive vision loss. From then on, I spent many nights in tears, searching endlessly for information. How effective would speech therapy be? Would he learn to walk normally? At what age would his vision start to fade? When would he go completely blind? How could he live independently then? I never imagined parenthood would begin with such heartbreaking questions.

On November 25, Yuge was taken into the operating room for a four-hour surgery to receive his cochlear implants. On December 10, his implants were activated. He remained calm, but he clearly reacted to sounds—that response gave us a moment of comfort in the midst of overwhelming sadness.

Over the following months, we returned to Geneva University Hospital twice a week for speech therapy and saw a physiotherapist for motor skills. Yuge worked so hard, making progress every single day. From not being able to lift his head or sit, he can now stand with support and even take small steps. From not responding to sounds, he now understands everyday conversations. To us, it feels nothing short of a miracle, and we cherish each moment of his growth.

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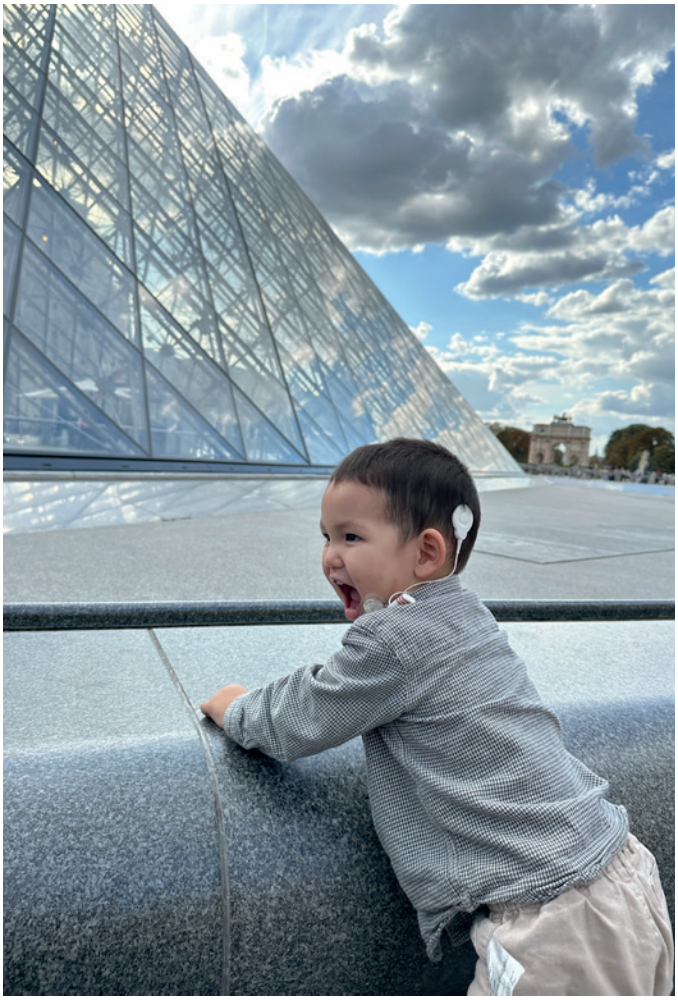
Yuge Continued from page 1

Now, more than seven months since his activation, Yuge can call us Mama and Papa. He loves imitating sounds, is endlessly curious about new things, also has developed his own little personality and hobbies. He enjoys climbing up and down slides and chairs, always trying to stand on his own. He greets people with a cheerful "hello" and "goodbye." He loves laughing, enjoys delicious food and music. To me, he is an angel baby.

Yes, watching him grow has eased, at least for now, the fears I carry for his future. I keep reminding myself not to let my worries steal the joy of his present. What we can do is stay informed, follow the latest research through the Usher 1F Collaborative, and share knowledge with the patient community we've built in China. Breakthroughs like David Corey's mini-gene therapy give us hope, and encouraging results from Nacuity's* clinical trials keep our spirits strong.

Yuge



Yuge enjoying his new home in Paris

In June, we took Yuge to the Usher Syndrome conference. Of course, he's too little to understand what this trip means, but we want to send him a message: face life with courage, live fully with Usher, and always hold on to hope. And he will never face these challenges alone—his parents will walk beside him every step of the way.

WE WANT TO SEND HIM A MESSAGE: FACE LIFE WITH COURAGE, LIVE FULLY WITH USHER, AND ALWAYS HOLD ON TO HOPE.

Our deepest wish is that Yuge will grow up to draw strength and wisdom from this unique life journey. May he make peace with himself, be strong and free, ordinary yet joyful. ◆

* Nacuity Pharmaceuticals shared promising clinical trial results showing that their experimental treatment, NACA, may help slow vision loss for people living with Usher syndrome.

BREAKING BARRIERS, BUILDING HOPE:

Usher 1F Research Takes Center Stage

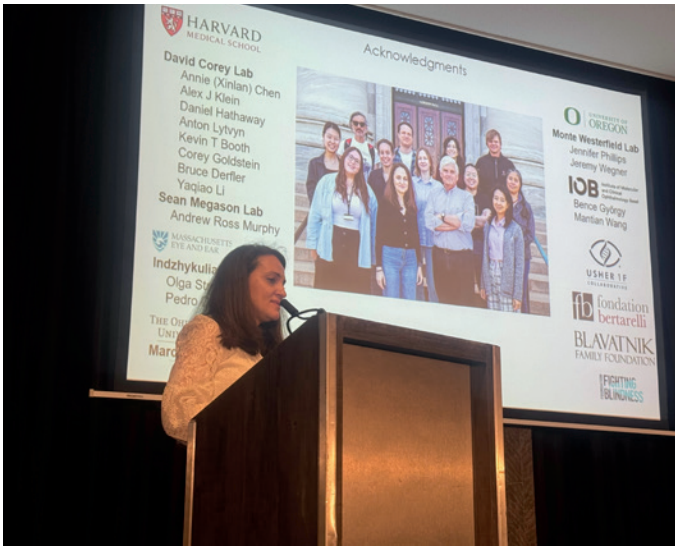
On June 19–20, research scientists and clinicians from around the world gathered in Nijmegen, the Netherlands, for the first in-person Usher syndrome scientific research conference since 2018. These meetings are invaluable, offering researchers the chance to step out of their silos, exchange insights, and explore how promising approaches for one subtype of Usher syndrome might be adapted to others.

A decade ago, not a single session at Usher conferences mentioned research toward a cure for Usher 1F. In 2025, however, we played a prominent role.

In her keynote lecture, Maryna Ivanchenko, MD, PhD, Harvard Medical School, presented her work in David Corey's lab on developing an Usher 1F mini-gene: *Developing Gene Therapies for Usher Syndrome Type 1F Deafness and Blindness*.

Another keynote was given by Marcos Sotomayor, PhD, University of Chicago, who trained as a postdoctoral fellow in David Corey's lab and was instrumental in the creation of our mini-gene. His lecture, *Zooming in on Usher Syndrome Proteins Cadherin-23 (USH1D) and Protocadherin-15 (USH1F)*, described his collaboration with David Corey in building the foundation for this critical work.

Jennifer Phillips, PhD, University of Oregon Institute of Neuroscience, delivered another keynote lecture, presenting her work in Monte Westerfield's lab: *Investigating General and Type-Specific Therapies for Usher Syndrome*, which included a focus on the lab's Usher 1F research.



Maryna Ivanchenko presents groundbreaking research advancing the search for an Usher 1F cure

Beyond learning from state-of-the-art scientific presentations, our cofounders Melissa and Elliot Chaikof connected with Usher syndrome families and leaders of other patient-led research organizations from both Europe and the United States during both the scientific research conference and at the family conference on the following day. In particular, they met two parents newly navigating an Usher 1F diagnosis and their adorable young son, Yuge, whose story we share in this newsletter.

The conference was an energizing and inspiring opportunity to both learn and contribute to the latest Usher syndrome research. We look forward with anticipation to the next gathering, hopefully much sooner than another seven years. ◆

Camaraderie among Usher families - Xin Chen with Yuge, Berta Adell, Melissa Chaikof, Chloe Joyner, and Jessica Chaikof

ESTIMATING IMPACT:

How Prevalence Research Strengthens Usher 1F Therapy Development

From 2020 to 2023, Usher 1F Collaborative benefitted greatly from being part of the first cohort of patient-led research organizations to receive a Chan Zuckerberg Initiative Rare As One Project grant. In addition to funding for capacity building, the grant provided extensive training on running a successful nonprofit and opened doors to opportunities that significantly advanced our mission.

One of the most valuable of these opportunities was engagement with the Rare Genomes Project at the Broad Institute of Harvard and MIT. Understanding the prevalence of a rare disease is critical - it influences researchers' resource allocation, guides scientific goals, and shapes the level of interest from biopharma companies. It also helps identify sub-populations that may be disproportionately affected by a genetic condition, such as those of Ashkenazi Jewish descent, East Asian heritage, and others.

Drawing on their expertise in genetic variant curation, the Rare Genomes Project team, using data from the Genome Aggregation Database (gnomAD), the world's largest human reference database developed at the Broad Institute, along with variants collected from additional databases, has been able to estimate the prevalence of Usher 1F and other rare diseases.

Our inclusion in this project enabled us to finally obtain prevalence numbers for Usher 1F:

PREVALENCE	
CONSERVATIVE*	**1 IN 1,720,051**
RELAXED**	**1 IN 382,103**

This corresponds to the following numbers:

NUMBER OF PERSONS WITH USHER 1F	UNITED STATES	WORLDWIDE
CONSERVATIVE	**199**	**4,786**
RELAXED	**896**	**21,535**

Because Usher 1F is a recessive genetic disease, meaning both parents must carry and pass on the mutation for their child to be affected, the number of individuals who carry an Usher 1F mutation is significantly higher than the number who actually have the disease.

NUMBER OF CARRIERS OF USHER 1F MUTATION	UNITED STATES	WORLDWIDE
CONSERVATIVE	**521,422**	**12,547,561**
RELAXED	**1,106,651**	**26,639,804**

These findings underscore the role of genetic testing in early diagnosis, family planning, and research.

While the Rare Genomes Project confirmed that Usher 1F is indeed an ultra-rare disease, their relaxed prevalence estimates suggest that more individuals are affected than we have yet been able to identify. This reinforces the importance of our ongoing outreach to medical professionals and educational centers to find and connect with additional people living with Usher 1F. Expanding this community is vital, not only to ensure that patients are aware of and can benefit from future clinical trials, but also to help attract the engagement and investment of biopharma companies. ◆

*Includes only genetic mutations definitely known to cause Usher 1F

** Includes variants of unknown significance, which are genetic changes that may cause Usher 1F but are not yet definitively classified as disease-causing

USHER SYNDROME GAINS RECOGNITION:

Dedicated ICD-10-CM Codes to Transform Care and Research

ICD-10 (International Classification of Diseases) codes are the worldwide standard for describing diagnoses, conditions, and reasons for health care visits. In the U.S., ICD-10-CM is the national standard for diagnoses.

Until now, **Usher syndrome** has never had its own ICD-10 code. Physicians had to classify hearing loss, vision loss, and balance problems separately, which obscured the true diagnosis. This left patients invisible in medical records and statistics, often struggling to get insurance coverage for needed care.

During our three-year Chan Zuckerberg Initiative Rare As One Project grant, we learned from other rare disease advocates the importance of specific ICD-10 codes. Partnering with the Usher Syndrome Coalition, the Usher Syndrome Society, and with the invaluable support of Drs. Rachel Huckfeldt, Eliot Shearer, and Margaret Kenna, Usher 1F Collaborative applied for a code. After three rounds of CDC hearings, we are thrilled to announce success: **a dedicated ICD-10-CM code for Usher syndrome, with subcodes for types 1, 2, and 3**.

Effective October 1, 2025, please make sure your doctors use these codes:

- **Q99811** Usher syndrome, type 1
- **Q99812** Usher syndrome, type 2
- **Q99813** Usher syndrome, type 3
- **Q99818** Other Usher syndrome
- **Q99819** Usher syndrome, unspecified

Why this matters

- **Recognition & legitimacy:** Usher syndrome will no longer be hidden under vague codes but will have formal recognition in the U.S. health care system.
- **Insurance coverage:** A precise code supports approval for medical visits, therapies, genetic testing, and assistive devices.
- **Coordinated care:** Because Usher syndrome affects hearing, vision, and balance, a single code ensures all specialists are documenting the same diagnosis, improving communication and treatment.
- **Research & clinical trials:** Patients can be identified more easily in health records, speeding recruitment for trials, which is critical as gene therapies move forward.
- **Public health & funding:** Federal and state agencies will be able to track prevalence and allocate resources, strengthening the case for research and patient support programs.

This is a milestone: a dedicated ICD-10-CM code finally gives Usher syndrome the recognition it deserves, paving the way for better care, access, and progress toward treatments and cures. ◆

Generosity in Full Swing at the 2nd Annual Golf, Tennis & Pickleball Outing

On June 9, supporters of Usher 1F Collaborative gathered at Mountain Ridge Country Club in West Caldwell, New Jersey, for the 2nd Annual Golf, Tennis & Pickleball Outing. The event also featured cards and mahjong, as well as brunch, a dinner reception with live music and an auction. Just like the previous year, guests opened their hearts and wallets, raising more than $320,000 for Usher 1F research.

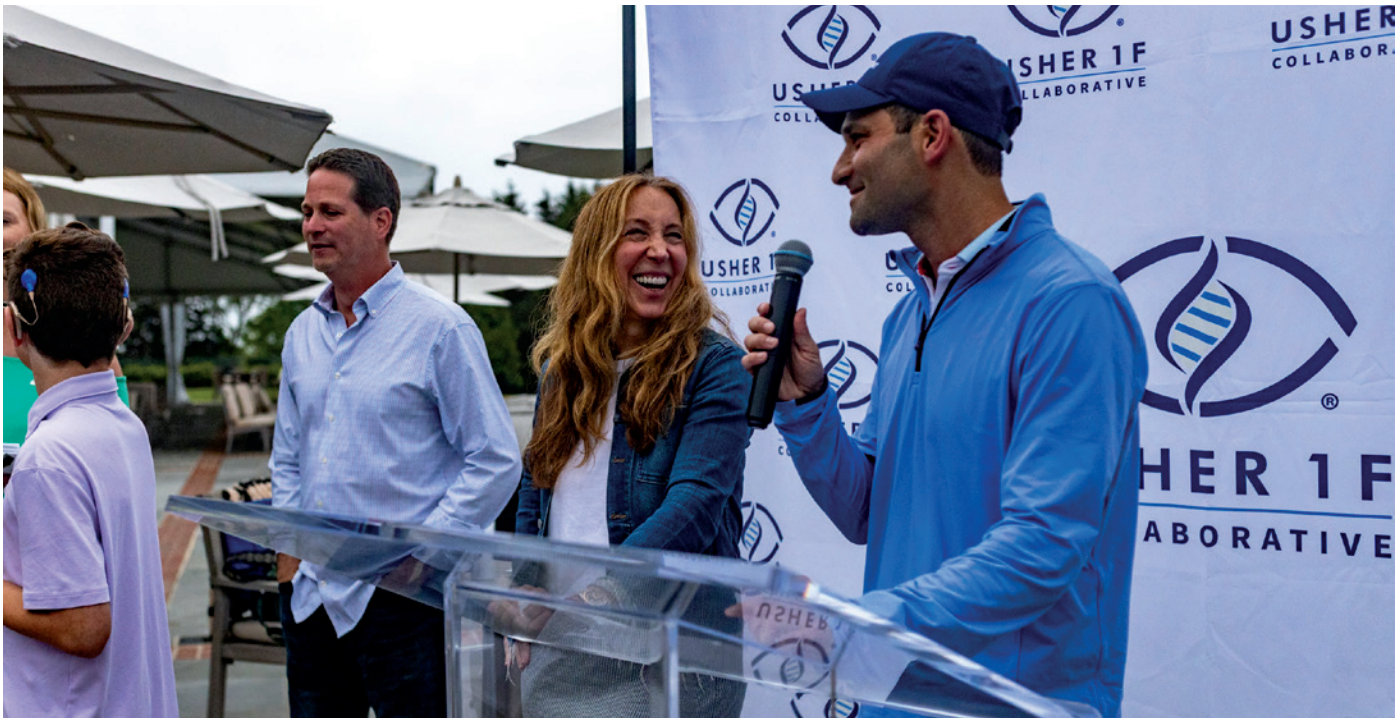
Although rain canceled the tennis and pickleball tournaments, the enthusiasm of the 250 attendees never wavered. The host committee remained the same for the event's second year, comprised of board members Josh Cohen, Eric Halper, Jared and Rachel Root, and Heather Rosenstein. Board member and Usher 1F Ambassador Dorie Shapiro attended, as well as Rachel Chaikof, who also lives with Usher 1F. Both offered inspiring remarks. Lead sponsors were Sarachek Law, I. Halper Paper, RBC, and Associated.

The evening concluded with a moving challenge from longtime donor Alan Fortier, who pledged to match additional contributions. Guests responded generously, adding even more momentum to the cause.

Mark your calendars: the 3rd Annual Outing returns on June 8, 2026. We hope to see you there! ◆

Golfers Mitchell Stein and Arthur Draznin enjoying the day

Zachary Root and his grandmother Anne Faust at a mahjong table



Board members Jared Root, Heather Rosenstein, and Eric Halper speak to the crowd

Turn Your Required Minimum Distribution into Hope for Families with Usher 1F

If you are 73 or older, the IRS requires you to take a Required Minimum Distribution (RMD) each year from your traditional IRA. These withdrawals are taxable as ordinary income, often creating a bigger tax bill than expected.

But there's a smarter way to meet this requirement: directing your RMD to Usher 1F Collaborative through a **Qualified Charitable Distribution (QCD)**.

Why Give Your RMD to Usher 1F Collaborative?

- **Reduce Your Tax Burden:** Donations sent directly from your IRA to Usher 1F are excluded from your taxable income.
- **Satisfy Your RMD Obligation:** Every dollar transferred counts toward your required distribution.
- **Accelerate the Search for a Cure:** Instead of paying more in taxes, your funds fuel groundbreaking medical research to treat and cure Usher 1F.

Turning your RMD into a gift ensures that what would have gone to taxes instead drives life-changing scientific progress and offers hope to families living with Usher 1F.

Another Smart Option: *Gifts of Stock*

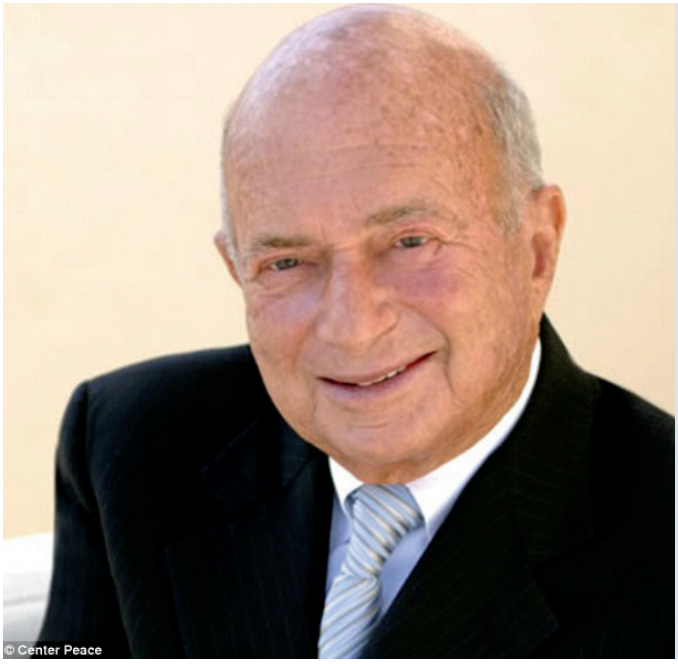
In addition to RMDs, donating appreciated stock is another powerful, tax-efficient way to support the Usher 1F Collaborative. By transferring stock directly, you avoid paying capital gains tax and may be eligible for a charitable deduction—while advancing the urgent mission to find a cure.

How to Give

- Contact your IRA custodian to request a Qualified Charitable Distribution to Usher 1F Collaborative (up to $100,000 annually can be given this way).
- Or speak with your broker about a direct stock transfer – we will provide you with Usher 1F Collaborative Fidelity account information.
- Let us know about your gift so we can thank you and ensure it goes straight to research.

Make Your Impact Today

Your RMD—or your stock donation—can change the trajectory of rare disease research. Together, we can speed the path to treatments and bring hope to every family affected by Usher 1F. ◆



In Memoriam, S. Daniel Abraham

With great sadness, we share the passing of S. Daniel "Danny" Abraham. In 2018 and 2019, Mr. Abraham quietly donated a total of $1 million to Usher 1F Collaborative, enabling our partnership with Foundation Fighting Blindness to conduct RUSH1F, a four-year natural history study of Usher 1F. This study, documenting the progression of vision loss in the absence of treatment, is providing critical data for future clinical trials. We are profoundly grateful for his generosity, care, and unwavering support, which have made a lasting impact on our mission.

Danny Abraham

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 **$1,323,905** $3,015,000

updated on September 17, 2025

usher1f.org/seeingforward

GIVINGTUESDAY

COMING SOON

DECEMBER 2, 2025