

# USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

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## **GROUNDING IN SCIENCE: August 2024**

A balance of research news and well-being for  
the Usher syndrome community

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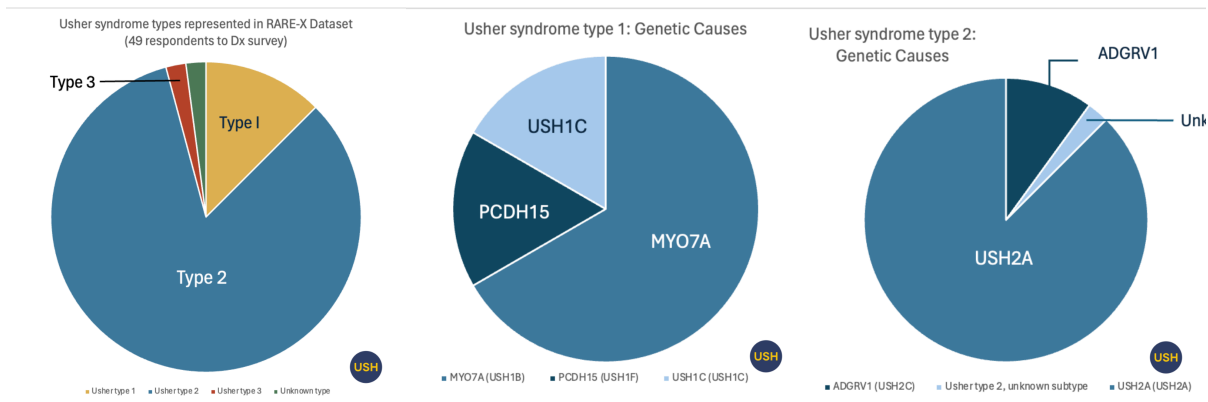
**USH Connections Conference | July 19-20, Rochester, NY + Online**  
**Hosted in partnership with the National Technical Institute for the Deaf**

We are still feeling so much gratitude and joy following the USH2024 Connections Conference. We'll soon share presentation recordings, transcripts, photos and more from this event - the largest gathering of the Usher community to date.

We are continually striving to expand and improve the USH Connections Conference. If you attended, please share your feedback on the #USH2024 Connections Conference here: [USH2024 Post-Event Survey](#).

### **Usher Syndrome Data Collection Program (USH DCP)**

Now is a good time to make sure you're in the [USH Trust](#) and [USH DCP](#). Once you've signed up for the USH DCP, make sure to fill out the diagnostic survey! We are trying to get 80 new signups by the end of the month so we can share more data like this:



For additional USH DCP sign-up support please [fill out this form](#).

## RUN FOR USHER SYNDROME

We are proud to announce that we are a charity partner with the Marine Corps Marathon, taking place on Sunday, October 27 near the Pentagon in Arlington, Virginia!

Are you hoping to run “The People’s Marathon” or know anyone who wants to run and needs a bib? 25 runners can claim their bibs and fundraise for the Usher Syndrome Coalition! [Sign up here](#) or email Sarah Jordan [scjord00@gmail.com](mailto:scjord00@gmail.com) with any questions!

**Deadline to sign up is August 31st.**

Have you joined the Usher Syndrome Coalition [Discord](#) Community Server? It’s a safe place for the community to connect with each other. Join here: <https://discord.gg/czwHGaDu7W>

## RESEARCH SPOTLIGHT

### ***Sepul Bio: Advancing Transformative RNA therapies for Inherited Retinal Diseases***

[Sepul Bio’s](#) team is excited to continue the work done at ProQR on the development of Ultevursen for the potential treatment of retinitis pigmentosa (RP) due to mutations in exon 13 of the USH2A gene.

As of December 2023, Sepul Bio, a new and innovative business unit of Laboratoires Théa, has taken on the clinical development of Sepofarsen for the treatment of Leber’s congenital amaurosis type 10, and Ultevursen for the treatment of USH2A mediated retinitis pigmentosa. This year will be a foundational year as the Sepul Bio team gets established.

The team, once established, will begin regulatory, clinical, and operational work with sites around the world. These sites are yet to be determined and will take time to become operational. Sepul Bio will communicate to the community once in a position to guide motivated future clinical trial participants to the appropriate clinical centers. The Sepul Bio team, notably Andrew Bolan, Patient Advocacy Director, who has transitioned over from ProQR and remains the throughline for this work, will update interested USH2A-related individuals and families once they have made further progress. Interested individuals can send inquiries to [contact@sepulbio.com](mailto:contact@sepulbio.com) and Andy will get back to you directly.

Ultevursen (formerly QR-421a) is an investigational RNA therapy that aims to restore vision in people with Usher syndrome and retinitis pigmentosa, or RP, due to mutation(s) in a specific part of the USH2A gene, called exon 13. If interested, it is recommended to make an appointment with your IRD or ophthalmology specialist to make sure all your tests and paperwork are up to date. It is also recommended to enquire with your specialist about eligibility due to the specificity of Ultevursen.

Speaking on the future development of Sepofarsen and Ultevursen, Jean-Frédéric Chibret, President of the Théa group outlined: *“For nearly 30 years, Théa has been committed to bringing the most modern and diverse range of innovative ophthalmic products to the market for the benefits of eye care practitioners and patients. We are very excited to continue the development of Sepofarsen and Ultevursen for patients. These two programs can deliver hope for patients suffering from retinal diseases that lead to blindness. We look forward to returning these assets into the clinic.”*

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Check out our Current USH Research page specific to [USH subtype](#) as well as [gene-independent therapeutic approaches](#).

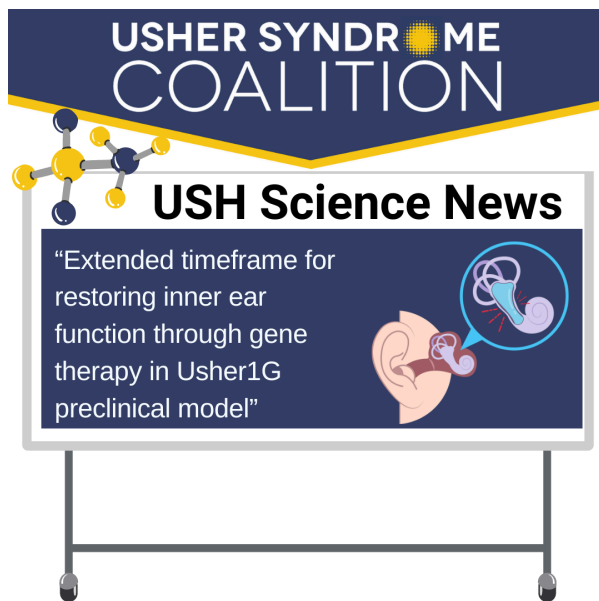
**[View Current USH Research](#)**

**IN CASE YOU MISSED IT: SCIENCE NEWS FEATURE**

***Extended timeframe for restoring inner ear function through gene therapy in Usher1G preclinical model***

January 9, 2024: Therapeutic research begins with testing new therapies in laboratory models of the disease before moving to human trials. However, translating success from models (cells or animals) to humans is challenging because they develop function and symptoms at different rates.

Recently, a team of researchers in Paris, France, successfully developed an adeno-associated viral (AAV) gene therapy that restored inner ear function in adult USH1G mice when the treatment was given just after birth. However, since all mice are born deaf and only begin to hear around the 2nd week of life while humans develop hearing in utero around the 19th week of pregnancy, it was unknown whether treatment after birth in humans (like was done in the mouse model), would work to restore hearing. To address this gap, the team expanded the treatment window for their USH1G mice. Their investigation revealed that the gene therapy could be administered for up to nine additional days, going beyond the mouse neonatal stage, while maintaining its effectiveness. This suggests administration of this gene therapy to newborn humans with USH1G instead of in utero may be possible.



### What this means for Usher syndrome:

As genetic testing becomes more common in Usher diagnosis, newborns with USH1G may have the opportunity to receive a gene therapy shortly after birth, potentially leading to enhanced vestibular function and hearing.

### READ ARTICLE

For more science news, check out our [Science News page](#), organized by treatment approach and type of Usher syndrome.

*DISCLAIMER: The Usher Syndrome Coalition does not provide medical advice nor promote treatment methods. USH Science News is intended to help summarize more complex literature for the community to use at their own discretion.*

## ON WELL-BEING: Post-Conference Emotions

Attending the Usher Syndrome Coalition's USH Connections Conference for the first time can evoke a wide range of emotions. Participants might feel hopeful as they connect with others who share the experience of living with Usher syndrome and learn about advancements in research and treatment development. There can be a deep sense of relief in finding a community where shared experiences help to ease feelings of isolation. Many may feel empowered by discovering advocacy resources and learning strategies to manage Usher syndrome, fostering a sense of control. Hearing success stories and witnessing the resilience within the community can be incredibly inspiring. The supportive atmosphere provides comfort and reassurance. However, the wealth of information and new experiences can also feel overwhelming. Ultimately, participants may leave with a profound sense of gratitude for the opportunity to connect, learn, and share, feeling encouraged by the dedication of researchers, advocates, and others committed to improving the lives of those with Usher syndrome.

Many people are told upon receiving an Usher syndrome diagnosis that they may never meet anyone like them. However, the USH Connections Conference defies these expectations, bringing together a vibrant and diverse community. At the conference, participants meet others who have lived with their diagnosis for their entire lives, as well as those who have just recently learned of it. They encounter individuals using white canes, guide dogs, sighted guides, co-navigators, spoken language, sign language, tactile sign language, protactile, and more. The conference exemplifies true accessibility, showcasing the wide range of experiences and communication methods within the Usher syndrome community.

It can be incredibly empowering to connect with others who truly understand your experience, who "get it." At the same time, it may raise questions about the future, such as, "What will my life be like?" or "What can I do now if there aren't yet any available treatments?" The experiences of someone who is culturally Deaf or DeafBlind may differ from those who have been encouraged to use spoken language. However, we all share a common goal of seeking connection and community, which is what makes these conferences so meaningful.

We are feeling a bit of the post-conference blues and wish we could still be with our USH Family. We'd like to use this time to remind you all of the mental health resources on our website: [Mental Health Resources](#). You will find different ways of connecting with the community, specific mental health support resources, presentations, and our previous "Grounded in Science Newsletters."

We share the research and peer-reviewed literature that offers insight into well-being: the science behind staying grounded. Fill out this [poll](#) to request a topic.

### Check out our Mental Health Resources webpage

*DISCLAIMER: The information and resources on this website are provided for educational and informational purposes only and do not provide medical or treatment advice.*

## USH Life Hack of the Day

Send your USH life hacks to [info@usher-syndrome.org](mailto:info@usher-syndrome.org)

Auracast is a new audio broadcasting feature that enhances Bluetooth technology. It allows devices such as hearing aids, cochlear implants, and other Bluetooth-enabled audio devices to receive clearer and more reliable audio streams over longer distances. This technology improves the overall listening experience by reducing latency and interference, making it especially beneficial for those with hearing impairments. Auracast can be used for streaming audio from TVs, smartphones, and other media sources directly to compatible hearing devices, ensuring better sound quality and convenience.

*While we share information on assistive hearing technology that may be beneficial, the Usher Syndrome Coalition does not endorse or promote any specific product.*



#### Our Contact Information

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