

# USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

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## GROUNDING IN SCIENCE: October 2024

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

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### **Staying Grounded in Science: Two Years of Progress and Growth**

Today, October 10, is World Mental Health Day. On this day two years ago, we promised to deliver featured science news and the latest progress in USH research, balanced with resources to support your well-being throughout this journey of understanding Usher syndrome.

When it comes to the abundance of research updates, take what serves you and leave the rest. In the meantime, we encourage you to make space for your well-being.

On the second anniversary of this Grounded in Science Newsletter, we thank you for being on this journey with us!

### **USH2024 Connections Conference**

Thank you to everyone who participated in the USH2024 Connections Conference, both in-person and virtually! We're proud to have hosted our largest conference ever, uniting the global Usher syndrome community.

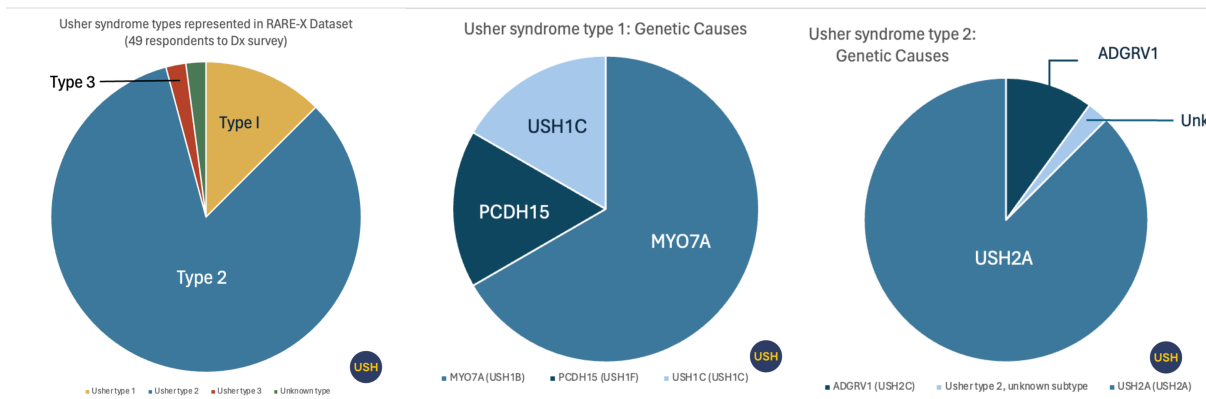
We welcomed 482 total participants from 20 countries - that includes 396 in-person attendees and 86 virtual attendees. We also worked with 92(!) Access Providers - interpreters, SSPs/CoNavigators, captioners, etc - meaning this gathering has touched a total of 574 lives!

Recordings and a recap of the USH2024 Connections Conference are [now available](#) for all to check out!

## Usher Syndrome Data Collection Program (USH DCP)

As the world continues to get to know the individuals living with Usher syndrome, it's a great time to join the [USH Trust](#) and [USH DCP](#) so researchers can better understand this diagnosis. If you'd like additional support enrolling, please [fill out this form](#).

Once you've signed up for the USH DCP, make sure to fill out the **Diagnosis Survey**, so you'll be counted in data like this:



## RESEARCH SPOTLIGHT

### ***AAVantgarde announces first patient dosed in First-In-Human Phase 1/2 LUCE-1 study, evaluating AAVB-081 (Dual-AAV) in retinitis pigmentosa related to Usher Syndrome type 1B***

[On September 16, 2024](#), AAVantgarde announced that the first participant received a dose in the Phase 1/2 LUCE-1 [clinical trial](#). AAVB-081 is an investigational gene therapy that is given via an injection in the back of the eye in the space just behind the retina (subretinally).

The goal of the LUCE-1 trial by [AAVantgarde](#) is to learn if AAVB-081, is safe to treat retinitis pigmentosa (RP) in participants with Usher syndrome type 1B (USH1B). USH1B is caused by mutations in the *MYO7A* gene. This trial will also look at how well the participants [tolerate](#) any potential [side effects](#).

Adeno-Associated Virus (AAV) is a small virus used as a vector - or a carrier - to deliver genes into cells. The *MYO7A* gene is too large to fit inside a standard AAV vector. AAVB-081 uses a proprietary “dual-AAV technology” (or two AAV vectors)

to deliver a functional copy of the *MYO7A* gene directly into retinal cells. Once at its final destination, the two parts are able to combine back into one whole working gene.

This process worked well in animals. This trial is the first to test this innovative approach in humans. Prof. Francesca Simonelli is leading the study. There will be multiple trial sites for LUCE-1 study. Learn more about AAVantgarde [here](#).

**What this means for the USH community:** The LUCE-1 trial represents a major advancement for the USH1B community, as it offers the potential for the “first ever” gene therapy treatment targeting progressive vision loss specifically for USH1B.

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

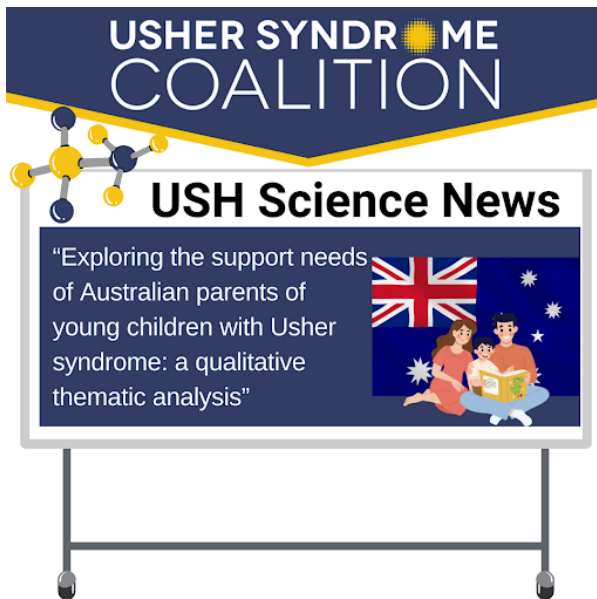
## View Current USH Research

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

#### ***Exploring the support needs of Australian parents of young children with Usher syndrome: a qualitative thematic analysis***

*March 21, 2024:* This study looked at the support needs of Australian parents with children who have Usher syndrome type 1. Researchers interviewed parents to learn about their experiences and needs.

**Four key support needs emerged:** social, informational, practical, and emotional. Social support was crucial, especially connecting with other parents in similar situations. Informational needs included accurate, timely diagnoses and detailed guidance from knowledgeable professionals. Practically, parents required respite care, case coordination, flexible work hours, and financial support. Emotionally, parents needed help coping with grief, managing their child’s progressive vision loss, and finding appropriate mental health support.



**What this means for the USH community:** The findings highlight the various ways parents need support when caring for children with this condition and can provide crucial insight for healthcare professionals and policy-makers.

**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. As always, consult with your trusted healthcare provider if you have questions or concerns about your situation.*

## ON WELL-BEING: Health Literacy

The U.S. Department of Health and Human Services (HHS) defines [personal health literacy](#) as the degree to which individuals can find, understand, and use information and services to inform health-related decisions and actions for themselves and others.

[Research shows](#) that hearing loss of any degree can affect the amount and quality of health information a person receives. This can impact both children and adults with Usher syndrome in several ways. First, they may miss out on casual family conversations, like learning about an aunt's breast cancer treatment or an uncle's recent diabetes diagnosis. Second, health-related articles and websites may not be accessible to individuals with low vision or those who rely on screen-reading technology. Lastly, the lack of sign language interpreters or other accommodations in healthcare settings can significantly limit access to information from providers, making it harder to understand one's own health history or feel empowered to advocate for oneself.

As clinical trial opportunities become available to the Usher community, health literacy becomes critical. The informed consent process can be overwhelming. Documents are often filled with complex medical and legal language even though research guidelines suggest that materials should be written at about a 6th-grade reading level. It's completely valid to feel unsure about what you're reading, and it's your right and responsibility to ask questions. Your medical team must do everything to ensure you understand before you consent to participate. You should also know that it is your right to stop participating in a study or clinical trial at any time. Make sure you understand all of the facts and the potential impact of stopping participation on your health and well-being.

The Coalition's science summaries and social media series are designed to help you build a strong foundation in the basics while breaking down complex concepts. In this way, you can feel empowered to ask critical questions, better understand the data, and make informed choices about your health.

Together, we can build a world where everyone has equal access to clear and useful health information, leading to better health outcomes. Visit [healthliteracymonth.org](http://healthliteracymonth.org) to learn how you can turn awareness into action!

### **Ways to improve your health literacy:**

- Find a medical provider who understands Usher syndrome and/or is willing to learn about your specific communication needs.
- Let your provider know your preferred language and communication methods (sign, spoken language, writing, apps, etc.). It is their responsibility to provide you access to clear speech, written materials, sign language interpreters or other supports as needed.
- Ask questions if something is unclear during your medical appointments. Your provider should take the time to explain.
- Speak up if you have concerns about your care or treatment.
- Ask for a written or electronic copy of the medical advice your doctor gives, and make sure you understand it fully before leaving your appointment.
- Choose well-respected and vetted sources for online health advice; not all sources are reliable.

### **References**

- Tolisano, A.M., Fang, L.B., Isaacson, B., Kutz, J.W., Hunter, J.B. [Can You Hear Me Now? The Impact of Hearing Loss on Patient Health Literacy.](#)

*Otology & Neurotology*, 41(8):p 1027-1032, September 2020. DOI:  
10.1097/MAO.0000000000002713

- Piao, Z., Lee, H., Mun, Y., et al. [Exploring the health literacy status of people with hearing impairment: a systematic review](#). *Arch Public Health*, 81, 206 (2023).
- National Institute of Health (NIH). [Health Literacy](#). 2021.

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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. Check out our mental health resources page on our [website](#). As always, consult with your trusted healthcare provider if you have questions or concerns about your situation.*

## USH Life Hack of the Day

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

One of our awesome USH Ambassadors recently got the chance to test out the new Meta AI glasses. Check out what she had to say about her experience!

"Ray-Ban Meta can make hands-free calls, send and receive messages, control music playback, provide real-time information like news, weather, and sports updates, and create content like stories, summarize documents and menus and more! Video calling capabilities are available through FB messenger and WhatsApp and are currently being beta tested in the Aira app to help users navigate in real time through video chat from a user's point-of-view.

As technology advances, so will the benefits of Meta AI in its ability to assist users who are blind and low vision. The speakers are nestled in the earpiece of the glasses and use open-ear audio technology that allows you to hear audio cues and responses, while still being aware of your surroundings. The audio feedback is discrete and prescription lenses are also available to order, making these glasses a helpful tool for the Usher community!"

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



**Our Contact Information**

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