

USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDING IN SCIENCE: October 2023

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

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Can you believe it's been a full year since we launched our Grounded In Science Newsletter? Last month we celebrated Usher Syndrome Awareness Day on September 16th. Did you see any of our [#USHDay2023 videos](#) that we shared on social media? Thank you to everyone who submitted video entries to be featured!

A couple announcements before we dig in:

1. In the U.S., you can get four free at-home COVID-19 tests this fall on [CovidTests.gov](#).
2. If you haven't checked your account on the [Usher Syndrome Data Collection Program](#) (USH DCP) lately, now is the time to do so! Be sure to upload your genetic reports and fill in the surveys as they appear on your portal. We will start compiling some of the de-identified data to share with the community.

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

GenSight Biologics

GenSight Biologics is a [gene therapy company](#) studying treatments for degenerative eye and central nervous system diseases. They are working on

using gene therapy, Mitochondrial Targeting Sequence (MTS), and optogenetics to develop therapies that can help prevent further degeneration in the eyes or even restore vision. Currently, [their GS030 product](#) is being studied for retinitis pigmentosa (RP). This product uses an AAV2 gene therapy vector to carry a photosensitive protein into cells.

In 2021, results from a study looking at GS030 in non-human primates were [published](#). The study results provided proof-of-concept of retinal ganglion cell activation through GS030 using optogenetic gene therapy. There was demonstrated pattern discrimination which was estimated to be 20/249 visual acuity. These results helped the company get approval for a Phase I/II clinical trial titled PIONEER.

PIONEER is an open-label dose escalation study looking at the safety and tolerability of GS030. In 2023, GenSight Biologics [announced](#) there were signs of efficacy and tolerability in participants of the PIONEER trial one year after getting GS030. Data collected from some participants followed for more than one year have also demonstrated the safety and tolerability of the product. The most common adverse event that was observed in the eye was intraocular inflammation that responded to corticosteroid treatment. With this information, the Data Safety Monitoring Board reviewing these data has recommended that the extension cohort (participants are currently being recruited) should receive the highest dose.

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

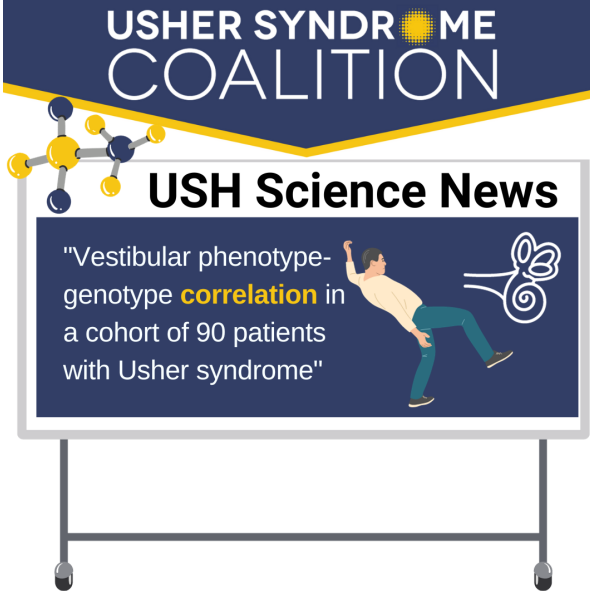
[View Current USH Research](#)

IN CASE YOU MISSED IT: SCIENCE NEWS FEATURE

Vestibular phenotype-genotype correlation in a cohort of 90 patients with Usher syndrome

November 3, 2020: In this study, researchers set out to understand the relationship between genes that cause Usher syndrome and vestibular dysfunction. It had been typically thought that those with USH1 and USH3 present with vestibular dysfunction in addition to the visual and hearing impairments. However, based on this study they noted that the gene identification alone is not enough to accurately predict the presence of a vestibular dysfunction. They

observed that individuals with USH2 also had vestibular dysfunction, even though most were asymptomatic and experienced normal balance.



The image shows a graphic for the Usher Syndrome Coalition. At the top is a dark blue banner with the text "USHER SYNDROME COALITION" in white, with a yellow sun-like icon between "SYNDROME" and "COALITION". Below this is a white banner with the text "USH Science News" in bold black. Underneath is a dark blue box containing the text: "Vestibular phenotype-genotype **correlation** in a cohort of 90 patients with Usher syndrome". To the right of the text is an illustration of a person in a white shirt and blue pants, appearing to be in motion or falling, with a white spiral graphic next to them. The entire graphic is supported by a grey stand with four wheels.

What this means for Usher

syndrome: Having more data with thorough documentation of the characteristics of Usher syndrome and how they are divided between the different types and atypical presentations of USH will allow clinicians to better diagnose and manage USH and researchers to better understand how to approach treatments.

[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: World Mental Health Day

October 10th is [World Mental Health Day](#), which is why we launched our Grounded in Science Newsletter on this day one year ago. This year, the theme is [mental health is a universal human right](#). In the spirit of this day, we invite you to explore our new compilation of resources for [Mental Health](#) on our website.

Usher syndrome (USH) is an unpredictable and progressive condition, which may impact your sense of identity across your lifespan - from being a hearing-sighted person to a person who is DeafBlind. ([Marine Arcous et al. 2020](#)) Due to the rare nature of USH, along with wide ranging communication modes, there is a lack of mental health professionals available to the DeafBlind community, forcing an already isolated population, those experiencing progressive hearing and/or vision loss, into further isolation without access to mental health support.

Facebook groups evolved as a way for adults diagnosed with Usher syndrome to provide peer support, and connect to others who “get it”; but facilitators living with this condition are already dealing with their own cycles of loss and grief, and have not always been formally trained in counseling, mental health, and life coaching.

For many years, the Coalition has recognized the need for more mental health resources for the Usher community. We’ve taken a small step towards building those resources by applying for and becoming one of the first grant recipients of the [RARE Mental Health Impact Grant from Global Genes](#). With this grant, we have conducted a “first-ever” 10-week training series for peer group facilitators who have Usher syndrome, led by Rebecca Alexander, psychotherapist with USH. This training focuses on establishing an accessible virtual environment, techniques of group facilitation, mediating conflict, navigating grief, maintaining neutrality, and resources for referral.

This 10-week training series empowers facilitators of USH Facebook groups and Discord chats, as well as our USH Ambassadors, to be a better resource for the community, following a “peer support model.”

The idea behind the **peer support model** is: “[Involving] a mutual exchange of practical and emotional support, based on ‘shared understanding, respect, and mutual empowerment between people in similar situations’ with critical ingredients such as shared responsibility, hope, self-determination over one’s life, and the use of lived experience knowledge” ([Smit et al. 2022](#)).

It is important to note that peer support ultimately doesn’t replace the years of professional training and experience held by a mental health professional.

Do you know the really great thing about the USH community? It’s filled with experts on Usher syndrome - **YOU**. You have the lived experience, which is invaluable, allowing you to have connections with peers who have been there before.

With your peers, it may be easier to feel a sense of connection and to create a safe place to share the various emotional ups and downs you experience. You might ask yourself what you feel when you receive unpleasant news. What thoughts and feelings do you have when you receive joyful news? With the support of peers with shared lived experiences, you become less isolated, and it becomes easier to be resilient as a community.

USH Life Hack of the Day

Send your USH life hacks to info@usher-syndrome.org

For those of you who enjoy hiking but feel that your traditional white cane isn't sturdy, consider checking out the **All Terrain Cane**.

"Whether you're taking a stroll down the block, at the beach, on snowy sidewalks, your grandchild's playground, or on the trails, the ATC gives you the power to explore where other mobility canes wouldn't dream to go." - [All Terrain Cane](#)



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