WWW.USHER-SYNDROME.ORG

USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: June 2024

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

Access the archives in English | Acceder al PDF en español

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

There are only 40 days until our next USH Connections Conference and **we need your help**. We are working hard with Rochester Institute of Technology (RIT) to find interpreters and CoNavigators for those who have requested them. **As our community continues to rapidly grow, we anticipate needing an unprecedented number of conference interpreters.** This requires us to widen our interpreter pool beyond Rochester.

We are reaching out to you, our USH community to ask for help. Will you donate to help cover the cost of interpreters coming from outside Rochester? We strongly believe in providing full access for everyone, and we can't do it without your help!

DONATE NOW

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

Odylia Therapeutics: Transforming Drug Development for Rare Disease

Odylia is a nonprofit biotech that supports rare diseases and drug development. It was initially formed with funding from the Usher 2020 Foundation and Mass Eye and Ear. The name Odylia is inspired by Odilia of Alsace, a patroness saint of good eyesight.

Many pharmaceutical companies overlook rare diseases with small patient populations due to the financial risk of bringing potential treatments to the clinic. This makes Odylia's nonprofit model an innovative strategy, emphasizing the importance of patient benefit and overcoming the traditional investment barriers for rare disease drug development.

In a traditional research model, scientists conduct research to discover and understand potential treatments, but they do not typically work on the later stages of getting these treatments to patients, such as clinical trials and commercialization. Pharmaceutical companies aim to develop and market treatments but they may be hesitant to invest in early-stage research that is high-risk, especially if the treatment may only help a small number of people or if profitability is uncertain.

That's where Odylia comes in. Odylia serves as a bridge between researchers and pharmaceutical companies. They help develop drugs and collaborate with academics and patient groups on their treatments. Their goal is to improve the likelihood of successful development and to facilitate partnerships with pharmaceutical companies to advance treatments to clinical trials. This model helps reduce the risks and challenges faced by researchers and pharmaceutical companies, potentially accelerating the development of new treatments and their availability to patients.

What this means for USH: As Usher syndrome is a rare disease, it has the potential of being overlooked by large biopharma companies who are concerned with profit margin. One of the projects currently in Odylia's pipeline is for a gene therapy for an USH1C vision loss treatment.

Check out our Current USH Research page specific to USH subtype as well as gene-independent therapeutic approaches.

View Current USH Research

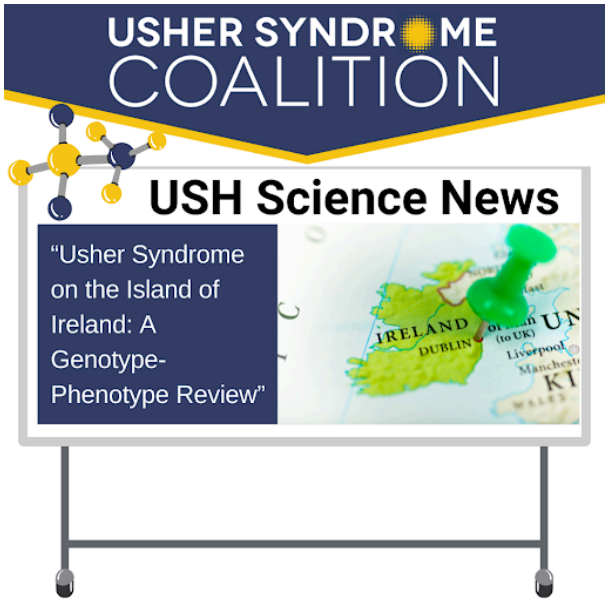
IN CASE YOU MISSED IT: SCIENCE NEWS FEATURE

Usher Syndrome on the Island of Ireland: A Genotype-Phenotype Review

June 3, 2023: Researchers studied Usher syndrome in Ireland to better understand the phenotypic and genotypic characteristics in this population.

145 individuals were identified from an Irish Inherited Retinal Dystrophy registry. In this cohort, Usher type 2 was the most common, followed by type 1, then type 3, and a single case of type 4.

Genetic testing, vision, hearing, and balance tests were measured. Of those with positive genetic testing results (82.1%), the most common genes were USH2A (causes Usher syndrome type 2a) and MYO7A (causes Usher syndrome type 1b). From the vision testing, the researchers found that the retinitis pigmentosa was similar in all types of Usher, differing only by age of onset and rate of progression.



What this means for Usher syndrome:

The similarity in the display of retinitis pigmentosa between all types of Usher syndrome in this cohort suggests that there is potential for gene-agnostic therapies that may benefit all types of Usher. Additionally, the distribution of genotypes in this study could be used to inform future clinical trials.

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: Movement

Hopefully, you are aware of the benefits of movement - how exercise is good for staying healthy, good for mental health, building community and self-esteem (Caretti et al. 2022). Maybe you feel limited, or unsure about how to make community sports or going to the gym more “Usher syndrome friendly.”

Jessalyn Akerman-Frank is a certified and trained Deaf yoga teacher who created a deafblind yoga class during the pandemic with adaptations, including “using ‘signs’ that represented the movement, props for tactile focus, and visualization techniques along with the sense of touch to maximize the experience.”

Going to the gym or a fitness class with a friend may help alleviate some of the anxiety of being in a dark, unfamiliar place. Working with the coaches to explain your hearing/vision needs helps them to better understand and make modifications that work for you. Using your cane, if you have one, can help you navigate spaces. Once you’re set up on a machine, it’s a great time to connect with yourself and be present in your body. Transitions may be challenging - it’s possible to have a system where the coach or instructor stands close to you while speaking or taps you on the shoulder to let you know. This can help you fully immerse in the group fitness experience.

Other activities like swimming can be enjoyable, whether you have waterproof hearing aids/cochlear implants, or you take them out to enjoy the silence. In a swimming pool, you can use the ropes separating each lane to guide you.

The loss of both hearing and vision can be disorienting for many reasons. Usher syndrome also affects postural stability - the ability to control the body's position in space to maintain balance and move - due to vision loss, and more so with vestibular dysfunction (Caldani et al. 2017).

A study in 2015 demonstrated the potential for a certain type of yoga to help visually impaired individuals develop somatosensory and vestibular responses, optimizing postural stability (Jeter et al., 2015).

Whatever your reason for prioritizing movement in your life, here are a few tips to help you feel more confident getting started:

1. Be patient with yourself- doing new things is hard and scary. It WILL get easier with time.
2. Don't be afraid of being uncomfortable- learn to laugh as you grow through the challenges.
3. Use your mobility & accessibility devices when possible and helpful.
4. Allow for clear communication before, during and after the activity to allow for feedback and ensure that your particular needs are being met.
5. Check out this document for more information.

How do you plan to move your body for mental health?

References:

Carretti G, Mirandola D, Sgambati E, Manetti M, Marini M. Survey on Psychological Well-Being and Quality of Life in Visually Impaired Individuals: Dancesport vs. Other Sound Input-Based Sports. *Int J Environ Res Public Health*. 2022;19(8):4438. Published 2022 Apr 7. doi:10.3390/ijerph19084438

Cavallo A, Ansuini C, Gori M, Tinti C, Tonelli A, Becchio C. Anticipatory action planning in blind and sighted individuals. *Sci Rep*. 2017;7:44617. Published 2017 Mar 17. doi:10.1038/srep44617

Rogge AK, Hamacher D, Cappagli G, et al. Balance, gait, and navigation performance are related to physical exercise in blind and visually impaired children and adolescents. *Exp Brain Res*. 2021;239(4):1111-1123. doi:10.1007/s00221-021-06038-3

Caldani S, Bucci MP, Tisné M, Audo I, Van Den Abbeele T, Wiener-Vacher S. Postural Instability in Subjects With Usher Syndrome. *Front Neurol*. 2019;10:830. Published 2019 Aug 8. doi:10.3389/fneur.2019.00830

Jeter PE, Haaz Moonaz S, Bittner AK, Dagnelie G. Ashtanga-Based Yoga Therapy Increases the Sensory Contribution to Postural Stability in Visually-Impaired Persons at Risk for Falls as Measured by the Wii Balance Board: A Pilot Randomized Controlled Trial. *PLoS One*. 2015;10(6):e0129646. Published 2015 Jun 24. doi:10.1371/journal.pone.0129646

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

One member in the community had the unique idea to put a glow collar, made up of glow sticks, on their black dog when letting them outside for potty breaks at night to help with visibility!

Our Contact Information
{{Organization Name}}
{{Organization Address}}
{{Organization Phone}}
{{Organization Website}}

{{Unsubscribe}}