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USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: March 2025

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

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Rare Disease Day is acknowledged worldwide on the last day of February to raise awareness and generate change for the 300 million people living with a rare disease, their families and carers.

Usher syndrome (USH) is a rare condition, impacting up to three major senses in the body: **vision, hearing, and balance.** USH is estimated to affect at least 25,000 people in the United States and over 400,000 worldwide.

Rare Disease Day emphasizes the power of community. **A rare disease brought a global community together. That's Usher syndrome.**

On this day, we hinted at something on the horizon: **Rare Genes**. Click here to see our Rare Disease Day post, a sneak peek of Rare Genes.

To our incredible community - stay tuned - this is for you.

RESEARCH SPOTLIGHT: NAC ATTACK

N-acetylcysteine, or NAC, is an oral tablet that the FDA initially approved as a new drug in 1963. There are many different applications for NAC, and in 1978, NAC received approval for use in treating Tylenol overdose. In April 2024, Johns Hopkins University launched Phase 3 of the NAC Attack clinical trial, which studies whether long-term NAC use can protect and maintain cone

photoreceptors (retinal cells responsible for color vision and visual acuity or sharpness) for those who have retinitis pigmentosa (RP).

The NAC Attack Phase 3 trial is expected to be a 45-month study, with goals to enroll approximately 438 patients across 30 sites in the US, Canada, Mexico, and Europe. Participants will be randomly assigned the treatment or a placebo. If NAC recipients show promising results at 21 months, the placebo group will also begin receiving the treatment.

This multi-center study is currently recruiting participants! If you are interested in participating, share your interest with your healthcare provider or retina specialist. They will confirm whether you qualify for the trial and help you with the next steps.

You can learn more about the trial from our 2024 USH Connections Conference in this video. Susie Trotochaud, Executive Director of the Usher 2020 Foundation and mother of two children with Usher syndrome, provides an overview of the NAC Attack trial.

Official trial information can be accessed here.

Participant criteria can be viewed here.

Did You Know? Did you know that the NAC Attack trial and a few other active clinical trials are also called basket trials? Basket trials describe clinical trials where more than one patient population can be studied together. In trials for gene-specific treatments, only patients with the specific genetic mutation of interest can participate. In NAC Attack, any individual with RP may participate regardless of gene mutation, as long as you meet the other criteria. Therefore, this ability to expand the patient population makes it a basket trial.

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

View Current USH Research

Clinical trials need participants. Let's make sure that there's a robust pool of potential participants when researchers are ready. Join the USH Trust today for the future of USH.

Join the USH Trust

IN CASE YOU MISSED IT: Science News Feature

Navigating the Usher Syndrome Genetic Landscape: An Evaluation of the Associations between Specific Genes and Quality Categories of Cochlear Implant Outcomes

February 26, 2024:
Patients with Usher syndrome, a genetic disorder that causes both hearing and vision impairment, often rely on accessibility tools to manage daily life. Common solutions for hearing impairment include hearing aids and cochlear implants (CI). Hearing aids, worn either in or behind the ear, amplify and transmit sound to the inner ear. However, for those with severe or profound hearing loss, hearing aids may be insufficient, making cochlear implants a better option.

Cochlear implants comprise two components: an external sound processor, which resembles a behind-the-ear hearing aid, and surgically implanted electrodes that transmit sound directly from the processor to the auditory nerve, bypassing the inner ear. While effective, cochlear implants are expensive, with costs ranging from $20,000 to $100,000, and require surgery. This led researchers at the Department of Audiology, Örebro University Hospital in Sweden to explore whether specific genes or genetic mutations could predict the success of cochlear implants.

In their study, which analyzed a database of Usher syndrome patients with known genetic mutations and cochlear implants, the researchers found that while most Usher-related genetic mutations did not correlate with CI outcomes, two genes did. Patients with mutations in the USH2A gene showed higher success rates with cochlear implants, while those with PCDH15 mutations tended to have poorer outcomes.

What this means for Usher syndrome: While using genetic mutations alone to predict cochlear implant success is overly simplistic and does not consider other variables such as the patient's health and age, surgical technique, and degree of hearing loss, this study shows how valuable genetic information can be in creating personalized treatment plans for patients with Usher syndrome.

Read Article

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.

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

ON WELL-BEING: Perseverance in the Face of Uncertainty: Living and Thriving with Usher Syndrome

Living with Usher syndrome presents daily challenges, but as Molly Watt shares in this patient perspective featured by MedJournal 360, it also fosters remarkable resilience. Diagnosed with Usher syndrome type 2A, Molly faced the gradual loss of vision and hearing, an experience that could have easily led to isolation. Instead, she embraced advocacy, technology, and community support to navigate life with confidence.

Molly's journey highlights the power of adaptability—learning to use assistive technology, advocating for accessibility, and refusing to let her diagnosis define her limits. Her story is a testament to the perseverance required to push through moments of uncertainty and the fortitude it takes to turn obstacles into opportunities.

If you're navigating life with Usher syndrome, know that resilience is already part of your story. Lean on your community, explore the tools that support you, and remember—you are not alone. Together, we strengthen one another, proving that perseverance can lead to empowerment.

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 Tip

Send your USH Tips to info@usher-syndrome.org

Hidden Disabilities Sunflower

The Hidden Disability Sunflower is for everyone with a hidden disability. Wearing the Sunflower is a way to signal that you have a disability or condition that may not be immediately apparent, and you may require some assistance or additional time. This is a great resource for traveling, especially in busy airports. The Sunflower is a symbol used internationally. You can select your home country on this website to view the global Sunflower network. There are businesses from every sector, and over 200 airports that recognize this symbol. If you prefer to have additional support from the Sunflower symbol, you can get a personalized ID card with more specific information regarding support you may prefer when asked and an emergency contact.

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