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USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: February 2025

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

Access the archives in English | Acceder al PDF en español

We want to assure you that the Usher Syndrome Coalition is working closely with other Usher syndrome organizations to monitor developments related to government and NIH funding and the potential impact on Usher syndrome research. As these funding decisions unfold, we remain committed to advocating for continued progress in research efforts that are critical to our community.

In addition to supporting research, ensuring accessibility for ALL individuals with Usher syndrome remains a top priority. We will continue to uphold and advocate for accessibility in all aspects of our work, ensuring that our community remains informed, empowered, and included.

We will keep you updated as we learn more and appreciate your ongoing support in advancing research, accessibility, and resources for individuals with Usher syndrome.

RESEARCH SPOTLIGHT: Nancuity Pharmaceuticals

Nacuity Pharmaceuticals, Inc., is a clinical-stage biopharmaceutical company developing treatments for retinitis pigmentosa, cataracts, and other diseases caused by oxidative stress. On January 21, 2025, they announced that its investigative therapy for the treatment of retinitis pigmentosa (RP), NPI-001, has received both **Fast Track Designation** and **Orphan Drug Designation** from the U.S. Food and Drug Administration (FDA).

NPI-001 is a unique form of N-acetylcysteine amide (NACA) tablets that help reduce oxidative stress, which is linked to RP and other diseases. Oxidative stress happens when there is an imbalance or too many naturally occurring free radicals in the body. In small amounts, free radicals can be beneficial; however, when they accumulate, they can harm our cells and contribute to disease progression.

Fast Track Designation is given to potential treatments that address serious or life-threatening conditions and meet unmet medical needs. This means that NP-001 can be reviewed faster and might get approved more quickly, so it can be available to patients sooner.

Orphan Drug Designation is awarded to drug candidates targeting rare diseases. This designation provides several benefits, like seven years of exclusive rights to sell the drug in the U.S., possible tax credits for eligible clinical trials, waivers for certain regulatory fees, and support to speed up the development process.

What this means for Usher syndrome: Currently, there is no FDA-approved treatment for all forms of RP. The only approved treatment for inherited retinal diseases (IRDs) is voretigene neparvovec (Luxturna), which is specifically for patients with ***RPE65*** gene mutations. Nacuity's NPI-001 gene-agnostic treatment has the potential to address a significant unmet medical need for all patients with RP, including Usher syndrome, no matter what their genetic mutations are.

[Link to Article]

Nacuity Pharmaceutical's Press Release

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

The USH3A causative gene clarin1 functions in Müller glia to maintain retinal photoreceptors

This study explores the role of clrn1 mutations in Usher syndrome type IIIA (USH3A). Using CRISPR/Cas9 technology in zebrafish, researchers deleted a large region of the clrn1 gene to mimic the mutation seen in USH3A patients. They found that clrn1 mutant zebrafish exhibited sensitivity to cell stress, age-dependent loss of photoreceptor function, and degeneration in the retina. Further investigation revealed disorganization in the outer retina, including abnormalities in Müller glia and photoreceptor cells. Re-expression of clrn1, specifically in Müller glia, prevented elevated cell death in mutant zebrafish exposed to high-intensity light, indicating a critical role of clarin1 protein in photoreceptor maintenance. Interestingly, high levels of clrn1 expression increased cell death in both wild-type and mutant animals, suggesting a need for controlled regulation.

However, re-expression of clrn1 in rod- or cone-specific cells did not fully rescue cell death. These findings suggest that clrn1 expression in Müller glia is crucial for photoreceptor support, possibly through direct interactions between Müller glia and photoreceptors mediated by Clarin1 protein.

What does this mean for the USH Community? This study's insights into the role of clrn1, particularly in Müller glia cells, shed light on the molecular mechanisms of vision loss in USH3A, potentially paving the way for targeted therapies, gene therapy approaches, and drug development to preserve photoreceptor function and slow disease progression.

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. RESILIENCE. FORTITUDE.

Living with Usher syndrome presents daily challenges and requires constant adaptation as we navigate our unique journeys through deafblindness. However, this diagnosis should never define us or limit our potential.
In the 2020 article *Living with Usher Syndrome: Patient and Physician Perspectives*, Ms. Helene Lønborg-Møller, a former psychiatrist, shares her

experience with Usher syndrome type 2A. Her story is a powerful testament to strength in the face of hardship. Over the years, she gradually lost both her sight and hearing, yet she never gave up. With each new challenge—from losing her vision to a more silent world as her hearing faded—she found ways to adapt and keep moving forward.

In her medical career, Ms. Lønborg-Møller changed specialties and moved several times to continue her work, even as her vision worsened. After the death of her husband, she faced the overwhelming pain of becoming completely blind and the isolation that comes with societal prejudice against the blind. Yet, she remained resilient, continuing to work, connect with others, and tackle life's difficulties.

Her journey is not just about survival but about living with purpose. She holds on to hope that new treatments may one day help our community regain sight. Through it all, she has persevered, reminding us that even in our darkest moments, we possess the strength to endure, adapt, and thrive.

This year, let's focus on **Resilience**, **Perseverance**, and **Fortitude** as key pillars of our well-being. As individuals living with Usher syndrome, we can embody these traits in our daily lives.

- **Resilience** – Every day, we adjust and adapt as our hearing and vision change. We find new ways to navigate our world, and even when things feel overwhelming, we keep going. Our ability to bounce back and continue moving forward is a powerful strength.
- **Perseverance** – We face challenges that others may not, but we keep working hard to maintain our independence, pursue our passions, and stay connected with the people around us. Even when the road is tough, we push through with determination and grit.
- **Fortitude** – We face the uncertainty of progressive hearing and vision loss. It requires courage to maintain a positive outlook and keep going. We confront life's toughest moments with the quiet strength that keeps us moving forward, no matter what.*t advice. Check out our mental health resources page on our website.*

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

Bluesky: Did you know you can also connect with us on Bluesky? We've noticed many researchers and scientists actively engaging there, and we look forward to connecting with you as well in this new space!
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