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USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: April 2025

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

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This month, we're excited to launch *Rare Genes*, a new campaign from the Usher Syndrome Coalition that reframes the narrative around Usher syndrome. Rooted in the rare genetic changes that connect our global community, *Rare Genes* celebrates the strength, resilience, and connection of those living with Usher syndrome. ***Rare Genes* brings our global community together.**

The *Rare Genes* campaign moves beyond the *medical model*—which focuses on diagnosing and treating a condition—and embraces the *social model*, which looks at how society can be more inclusive and accessible. It highlights the power of community and empowerment, showing what's possible even in a world not always built for us.

We've long worked to make science news accessible through clear, plain language—because we believe everyone should be able to understand and engage with the science that affects their lives. Now, with Rare Genes, we're leveling up our science education series to bring even more engaging and understandable content to our community. Stay tuned for what's next!

RESEARCH SPOTLIGHT: OCUGEN RECEIVES FDA APPROVAL FOR EXPANDED ACCESS PROGRAM

Ocugen, Inc., is working on a new potential treatment for retinitis pigmentosa (RP), an eye disease that causes vision loss over time. Their gene therapy, OCU400, is a possible one-time treatment being studied in the liMeliGhT trial.

After promising results in early studies (Phase 1/2), they are now in Phase 3, the last step before the company can apply for approval from the U.S. Food and Drug Administration (FDA).

Usually, people can try new treatments only by joining a clinical trial or waiting until the treatment is fully approved, which can take years. The liMeliGhT trial was initially designed only for people with RP caused by a change in the *RHO* gene. Through the Expanded Access Program (EAP), the FDA is letting Ocugen give OCU400 to up to 75 people with RP regardless of their genetic changes. To qualify, patients must still have some healthy photoreceptors left, even if they are in the advanced stages of RP.

What this means for Usher syndrome: Since RP is part of Usher syndrome, this program means all individuals 18 and older can be considered for the OCU400 gene therapy as long as they meet the necessary conditions.

Read Ocugen's press release

View OCU400 Clinical Trials page

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

In late 2024, we launched our new Science Education social media campaign to help simplify and explain the science behind this complex genetic disorder that affects our community. Last month, we introduced a new series on Gene Therapies - what it is, how it works, and how it is given to patients. Our community showed a lot of interest. For those of you who are not on social media, we wanted to share it with you here.

Gene-Based Therapies for Usher syndrome and RP:
Scientists are making tremendous progress using gene therapy to treat inherited eye diseases like Usher syndrome and retinitis pigmentosa. This month, we will explore gene therapy—what it is, how it works, and how it is given to patients.

Genes are DNA fragments that act like instruction manuals for cells. They tell cells how to make proteins, which help different parts of the body work. In Usher syndrome, specific genes have mutations, or changes, that stop the body from producing the proteins needed for hearing and vision.

People with Usher syndrome usually have mutations in both copies of a gene. These mutations can either create a faulty protein that doesn't work or stop the body from making the protein at all. Since these proteins are important for vision, their absence causes the light-sensing cells in the eye (photoreceptors) to break down over time, leading to vision loss.

Gene therapy is a way to help cells make these important proteins again. There are two main types:

1. **Gene Editing**: Fixes one or both mutations in the patient's gene.
2. **Gene Replacement**: Adds a new, healthy copy of the gene.

Next in the series: **Gene Therapies: How Do We Deliver Them to the Retina?**

Check out the blog series on Gene Therapies

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:
Internalized Ableism, Advocacy and Mental Health

By Becca Brown, LCMHCA, North Carolina USH Ambassador

The intersection of disability, identity and mental health is something near and dear to me. As a mental health provider, I work with a diverse population, focusing on disability, including neurodiversity, sensory loss, intellectual disability and chronic health conditions. Prior to becoming a therapist, I worked in the developmental disability residential field. I received a master's degree in Disability Studies in New York during a time when I identified myself as hard-of-hearing. About a year before I went back to school to become a mental health therapist, I learned I had Usher syndrome 2a. Having learned this new aspect of myself, I was suddenly contending with a new identity and meaning of disability as it shows up in my life.

My own journey with therapy has helped me understand what it means to advocate for myself; when to disclose and how to unpack feelings of shame, driven by my own internalizing of ableism. Internalized ableism, simply put, is when disabled people perceive and internalize society's negative views on disability. This can lead to intense feelings of shame, low self-esteem and hopelessness (Johannsdottir et al., 2022). These mental health concerns can lead individuals to fearing asking for help or changes in environmental supports, fears of disclosure or masking (Kidwell et al., 2023). My own journey, not only contending with hearing loss, but the added loss of visual input, has placed me in several situations when I not only want to ask for help, but NEED to ask for help. I've learned to advocate for myself not through grandiose means, but rather, by asking for what I need, such as, "Hey, when we go to that concert, could you help me navigate?" Or while playing cards, "Could we turn the lights up in here?"

Feelings of grief and loss while processing this diagnosis is ongoing. Acceptance may be a by-product, however, acknowledging the loss and change can help move one towards resilience, perseverance, and fortitude. Seeking a mental health therapist who is well versed in disability can be life-changing. Also, the more you can connect with others who are experiencing what you are going through is really important – I have been so grateful for my own connection to other mothers with Usher syndrome. There are several support groups on platforms such as Facebook and Discord. Knowing that you are not alone - and others can commiserate with the on-going loss, sadness and small victories, can be monumental.

Becca lives in North Carolina with her partner and two kids. She enjoys working in the garden, running and hula-hooping. She owns a private practice called Vital Minds Counseling and Services PLLC.

References

Johannsdottir, A., Egilson, S. P., & Haraldsdottir, F. (2022). Implications of internalised ableism for the health and wellbeing of disabled young people. Sociology of Health & Illness, 44(2). 360-376, doi:10.1111/1467-9566.13425

Kidwell, K. E., Clancy, R. L., & Fisher, G. G. (2023). The devil you know versus the devil you don't: Disclosure versus masking in the workplace. Industrial and Organizational Psychology, 16(1). 55-60, doi:10.1017/iop.2022.101

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.

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

USH Tip

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

USH Ambassador Jon Schultz loves his portable cordless table lamp that he brings to restaurants (some restaurants already have them). It makes it easy to read the menu and enjoy your food in a dimly lit environment. There are many options to choose from.

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