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USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: May 2025

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

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May is Mental Health Awareness Month. This is a time to raise awareness, foster open conversations, and reduce the stigma surrounding mental health. For the Usher syndrome community, the challenges of managing symptoms, navigating inaccessibility, coping with overstimulation, and processing emotions can all have a significant impact on our well-being. In this month's *On Well-Being*, guest writer Rebecca Alexander shares a powerful piece on how an Usher syndrome diagnosis can affect mental health, along with practical tools and strategies for support.

USHER SYNDROME DATA COLLECTION PROGRAM

As the world continues to get to know the individuals living with Usher syndrome, it's a great time to join the Usher Syndrome Data Collection Program - the USH DCP - so researchers can better understand this diagnosis. If you'd like additional support enrolling, please reach out to Yael Saperstein, our Community Enrollment Coordinator for the USH DCP. Yael is an expert on the enrollment process, accessibility, and guiding new participants every step of the way. Contact Yael here: y.saperstein@usher-syndrome.org.

RESEARCH SPOTLIGHT: jCYTE, INC.

Clinical Trial Announcement!

jCyte, Inc. is a biotechnology company based in California. They are developing a

new treatment called jCell, a cell therapy that may help people with retinitis pigmentosa (RP). This potential treatment is gene-independent, meaning it could work for people with any form of RP, no matter what gene caused it. On April 22, 2025, jCyte announced they are now recruiting adults with RP for their JC02-88 Phase 2 clinical trial.

What is jCell?
jCell is a cell therapy made from living cells. It is designed to help protect and possibly improve vision. The therapy is given through a one-time injection into the vitreous, or jelly-like substance in the middle of the eye.

Study Overview:

1. Enrollment goal: Up to 60 people with RP
2. Treatment: A single shot of jCell (with 8.8 million cells) or a sham procedure
3. Duration: 6 months
4. Location: Southern California, USA: Irvine, Beverly Hills, and San Diego, CA

Who Can Join?
To join, you must:

1. Be 18-60 years old
2. Able to travel to Southern California for all study visits during the 6-month study period
3. Have a confirmed diagnosis of retinitis pigmentosa
4. Have vision between 20/80 and 20/800 in one eye
5. Still have at least 8° central vision in the same eye where you can see clearly
6. No previous jCyte study participation
7. No trial participation in the last 6 months, except for NAC trials

Learn More:
jCyte Study Page: Click Here
Clinical Trials Page: Click Here

Interested in joining?
Talk with your retinal specialist. They can help you determine whether you qualify and explain the next steps.

If you have further questions, you can contact jCyte at info@jcyte.com.

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

Elasticity and Thermal Stability are Key Determinants of Hearing Rescue by Mini-Protocadherin-15 Proteins

June 17, 2024: USH1F is caused by mutations in the protein Protocadherin-15 (PCDH15). Because PCDH15 is a very large protein, the genetic sequence that cells use to make PCDH15 protein is too long to fit into the viruses that are used to deliver gene therapy treatments.

In a previous study, these researchers tried to overcome this "size" problem by examining the PCDH15 protein and designing smaller versions of it (called mini-PCDH15). They hoped these mini-PCDH15s would accomplish the same function as the regular large version of PCDH15. When the researchers used a virus to deliver their different mini-PCDH15 proteins to mice with mutant PCDH15 and hearing loss, they improved hearing function. However, they found that some of their mini-PCDH15 proteins worked better than others, and they were unsure why.

In this study, they wanted to identify why some mini-PCDH15 proteins work better than others at restoring hearing in mice with mutant PCDH15. They selected three of the mini-PCDH15 proteins they had tested in mice previously, two of which restored hearing function in mice and one that did not, and they did experiments to try to identify what was unique about the mini-PCDH15 proteins that worked well. They found that one of the biggest differences between the mini-PCDH15 proteins that worked and those that did not was their stability at higher temperatures and their flexibility. The best mini-PCDH15 protein was flexible and stable at higher temperatures, whereas the worst mini-PCDH15 was rigid and less stable at high temperatures.

What this means for Usher syndrome: By understanding what characteristics are important to maintain as a protein is shortened, researchers will be able to design better versions of mini-PCDH15 for gene therapies in the future. In addition to PCDH15, many other proteins involved in Usher syndrome are also very large. The viruses used for gene therapies cannot deliver their full-length genetic sequences. So, this same “mini-protein” strategy could be used to overcome this problem.

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:
Living With Usher Syndrome: Practicing, Prioritizing, and Protecting Your Mental Health

By Rebecca Alexander, LCSW-R, MPH, RYT, PLLC

Getting an Usher syndrome diagnosis isn’t a single, life-altering moment - it’s a process. A series of reckonings. A gradual evolution of internal and external shifts. Some are quiet and subtle; others hit hard and feel gut-wrenchingly sad. What’s certain is that it’s never just about your vision or hearing. It’s about your identity, your independence, your relationships (especially with yourself), your livelihood, your confidence, your future, and your sense of safety in the world.

My diagnosis didn’t arrive all at once - it unfolded in stages. Genetic testing and early screening have come a long way since then, but when I was told I would lose both my vision *and* hearing, I couldn’t wrap my head around it. None of us can. The reality settled in slowly - struggling to read a menu in dim light, hesitating at a busy crosswalk, missing a punchline in a loud room. The world became harder to navigate before my emotions could catch up.

As a psychotherapist, I understand that grief doesn't wait until after the loss. It's there with you while you're still functioning, still showing up, still doing your best. That's what makes living with Usher syndrome so complex. You're adjusting to what's slipping away, trying to stay grounded in what remains, and bracing for what's next. It's a constant balancing act between presence and preparation, acceptance and anticipation.

There's no single right way to cope, but these practices have been essential for me:

- **Make room for all of it.** Gratitude and grief often coexist. You can be thankful for what you have and still mourn what's changing or what you've lost. That's not a contradiction - it's what makes us human. The more space you allow for the full range of emotions, the more compassionate your relationship with yourself will become.
- **Let people in. An Usher syndrome diagnosis won't break your spirit - but isolation can.** Whether it's a therapist, a trusted friend, or someone who just "gets it," connection is essential. You don't have to do this alone. Support reshapes how we cope.
- **Name what you feel and what you need.** The more clearly you can express your experience, the easier it is for others to support you - and for you to stay connected to yourself. Saying, "I'm doing my best, but this is really hard," or "I'd love to join, but I'll need a ride," doesn't make you a burden. It means you're honest, and it allows others to connect with you.
- **Practice non-judgmental self-awareness.** Notice what you're feeling without rushing to fix it, numb it, or push it away. Some days are just hard. That's okay. Let it be true.
- **Come back to your breath.**
  Close your eyes or lower your gaze. Place one hand on your chest or belly.
  Feel the gentle expansion and release of your breath.
  You don't need to change anything - just notice.
  Let your breath anchor you in the present moment. This is your home base.
- **Give yourself time to grieve.** Grief doesn't define you - it makes space for joy, connection, and wholeness. Mourning what's changed and what may still change is not weakness - it's part of being deeply human.

About Rebecca Alexander:
As an adolescent, Rebecca received a life-changing diagnosis: retinitis pigmentosa (RP), a rare genetic condition that would gradually narrow her sight until she was completely blind, likely by age 30. At 19, still recovering from a fall

that left her athletic body shattered, Rebecca was diagnosed with Usher syndrome, the leading genetic cause of combined blindness and deafness.

Now in her 40s, living with a sliver of sight and total deafness without cochlear implants, Rebecca faces life with unwavering determination and a deep commitment to connection. Through her journey, she proves that life's greatest challenges can open the door to profound strength and a deeper sense of self.

With two Master's degrees from Columbia University and advanced certifications, Rebecca has a thriving psychotherapy practice in Manhattan. Rebecca also created and facilitates an in-depth mental health curriculum for mental health professionals as well as for organizations.

To learn more about Rebecca, visit her website here or follow her on Instagram here.

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

With warmer months approaching in the Northern Hemisphere, sun protection is important for longer days and more sun exposure. Polarized sunglasses are a great option. They deflect UV rays away from your eyes while reducing glare and improving contrast. Polarized lenses help reduce eye strain in bright, sunny outdoor settings.

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