

# USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

## GROUNDING IN SCIENCE: November 2023

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

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Happy November! This is a month many associate with gratitude and thankfulness. We at the Usher Syndrome Coalition are deeply appreciative to have you as a cherished member of our ever-expanding global Usher community.

In case you missed it, we recently shared an exciting update: the USH2024 Connections Conference is just around the corner! This gathering will take place in Rochester, New York, offering an enriching experience for those who can join us in person. For those who prefer the convenience of attending online, the conference will also be live-streamed, ensuring you can participate from the comfort of your own space.

USH2024 participants have the choice to [register for IN-PERSON or VIRTUAL access here](#). To get a sense of what you might expect at the USH2024 event, you can check out the [USH2022 recap and recordings](#).

Thank you for being a vital part of our community. We look forward to connecting with you at this remarkable event and continuing our journey together.

*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

## ***NAC Attack***

[Johns Hopkins University](#) is recruiting for “[NAC Attack](#), A Multicenter Placebo-Controlled Clinical Trial to Test Oral N-Acetylcysteine (NAC) in Patients with Retinitis Pigmentosa (RP).” This study will test whether taking NAC long-term can slow the progression of vision loss in a person with RP. N-acetylcysteine is a strong antioxidant that is FDA-approved for acetaminophen (paracetamol) overdose.

NAC is being explored as a potential treatment approach for retinitis pigmentosa. In RP, mutations -or mistakes in the genetic code- cause degeneration of rod photoreceptors which are responsible for vision in dim lighting, resulting in night blindness. After rod photoreceptors are eliminated, gradual degeneration of cone photoreceptors occurs, gradually narrowing peripheral vision and eventually causing tunnel vision. [Oxidative stress](#) contributes to cone degeneration and, ultimately, loss of vision. NAC has been shown to reduce oxidative stress, and orally administered NAC in a mouse model of RP slowed cone degeneration.

In a [phase I clinical trial](#), patients with RP were given NAC orally for 6 months; it was well-tolerated and even indicated small improvements in visual acuity in the retina. This suggests that long-term administration of NAC may promote survival and maintenance of cone function in RP. NAC Attack is a phase III, multicenter, randomized, placebo-controlled trial that will determine if oral NAC provides benefit and is safe in patients with RP.

[Watch](#) Dr. Peter Campochiaro’s presentation from the USH2022 Connections Conference to learn more about this clinical trial:

## NAC Attack

Multicenter randomized placebo-controlled trial  
Designed to determine if long-term NAC slows progression of RP and is safe

30 centers in US, Canada, Mexico, and Europe

438 patients

Study duration = 45 months

7 In-person visits: every 9 months + M0 + M40.5

3 Tele-visits: M13.5, M22.5, M31.5

10 Phone calls: M2.25, M6.75, M11.25, M15.75, M20.24, M24.75, M28.25, M33.75, M38.25, M42.75



**said they would be enthusiastic  
about participating**


For more, 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 proteins ADGRV1 (USH2C) and CIB2 (USH1J) interact and share a common interactome containing TRiC/CCT-BBS chaperonins***

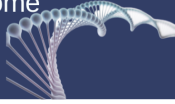
*June 22, 2023:* Mutations in the genes CIB2 and ADGRV1 have been described as causative for Usher syndrome subtypes USH1J and USH2C, respectively. The researchers set out to identify how these proteins function and found that these proteins work together in common pathways. Further investigation showed these proteins interact together in light-sensitive cells in the retina.



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**USH Science News**

“Usher syndrome proteins ADGRV1 (**USH2C**) and CIB2 (**USH1J**) interact and share a common interactome containing TRiC/CCT-BBS chaperonins”



### What this means for Usher

**syndrome:** This study expands our understanding of the function of molecules associated with Usher syndrome. This information may help researchers identify possible targets for the development of treatments.

[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: Meet the Team!

As we celebrate a year of Grounded in Science Newsletters, we recognize that we touch on tough topics monthly. In the interest of transparency, we want to introduce you to the team behind the Grounded in Science and Well-Being series. This group, from diverse backgrounds, reports relevant research to the community.



[Meet Lawreen Asuncion](#): Lawreen graduated with a Bachelor's Degree in Biological Sciences and has worked professionally in the biotech and life science markets for over 25 years. Having Usher syndrome type 2C, Lawreen is passionate about fostering awareness around Usher syndrome and supporting research for rare genetic diseases.



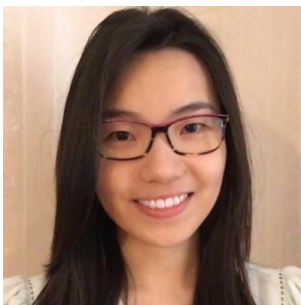
[Meet Rya Freeland-Kiablick, MSW](#): Rya graduated with a Master's in Clinical Social Work from Boston College in 2013 and has worked as a mental health clinician for individuals throughout their lifespan. She is a mother of a child with Usher Type 1 and provides context from both a personal point of view and a professional point of view. "It is important for me to be involved with the Usher community because it provides my family with support as well as a greater awareness of experience and needs. As a social worker and a parent, I am an advocate, and I understand the importance of addressing the topic of mental health within this community."



[Meet Monica Pruitt](#): Monica graduated with a Bachelor of Science in Biochemistry and Molecular Biology and has Usher syndrome type 2. "I write speaking from my own experience coping with the diagnosis as well as what I've observed in the community." Combining her technical science training with her personal mental health journey makes her a well-rounded writer, writing from experience and backing it with peer-reviewed journal references.



**Meet Lisa Rowinski**: Our Well-Being series copy editor, Lisa Rowinski, NIC, MA, is a nationally certified ASL interpreter with a Master's Degree in Counseling. Lisa has worked with folks in the USH community in a variety of capacities, from case management to interpreting, to counseling. Lisa has a special interest in the DeafBlind community and continues to read, attend workshops, and study the mental health and linguistic/communication needs of this diverse community. Lisa is also a certified Pilates instructor and believes that movement of any kind is a great benefit to everyone's physical and mental health!



[Meet Kayleigh To](#): Kayleigh graduated with a Master's in Public Health and has a strong interest in health policy. She interned at the Usher Syndrome Coalition before moving on to a volunteer position. Her work at the Coalition has driven Kayleigh to be eager to spread awareness and help out the Usher community in any way she can.



[Meet Krista Vasi](#): Krista graduated with a Master's in Public Administration, with a focus on nonprofit management, from the Maxwell School of Syracuse University in 2008. As the Usher Syndrome Coalition's Executive Director, Krista manages the Grounded in Science News Committee and the Well-Being series and is an editor for this newsletter.

It's valid to feel nervous or uneasy when talking about mental health. But the reality is it helps more than it hurts. As our beloved "neighbor," Mister Fred Rogers once said:

"Anything that's human is mentionable, and anything that is mentionable can be more manageable. When we can talk about our feelings, they become less overwhelming, less upsetting, and less scary."

“**Anything that's human is mentionable, and anything that is mentionable can be more manageable. When we can talk about our feelings, they become less overwhelming, less upsetting, and less scary.**”- Mr. Fred Rogers

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## USH Life Hack of the Day

Send your USH life hacks to [info@usher-syndrome.org](mailto:info@usher-syndrome.org)

“Learn how to type on your computer without needing to look at the keys. This makes it easier to follow what you are writing on the screen (or with TalkBack/VoiceOver/screenreaders) and prevent neck strain from having to look up and down frequently.”- Monica P, USH2A



### Our Contact Information

\*{{Organization Name}}\*  
\*{{Organization Address}}\*  
\*{{Organization Phone}}\*  
\*{{Organization Website}}\*

\*{{Unsubscribe}}\*

