

# USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

## GROUNDING IN SCIENCE: December 2024

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

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As we close out another remarkable year, we're deeply grateful for the community that drives our mission forward. Together, we advance scientific progress by fostering meaningful connections between researchers and families. Research toward treatments depends on an active and engaged community of participants. In short, **Research + Community = Treatments**—and you make this equation possible.

For those looking to deepen their impact in 2025, there's a special opportunity to [join our USH Visionaries](#)—a dedicated group of monthly donors fueling our efforts to build an engaged and empowered community.

When you become an USH Visionary at \$50 or more per month, you'll receive *Sunrise*, the second painting in the stunning "USH Steeple Study" series by master painter Timothy Chambers. This limited series captures the evolving view of a steeple through the lens of someone with retinitis pigmentosa. USH Visionaries who joined last year received *Dawn*, and this year, you'll have the chance to begin or continue your collection as a heartfelt thank you for your support.



Now, let's dive into this month's balance of research progress and resources to support your well-being.

## RESEARCH SPOTLIGHT

### ***Sepul Bio announces first participant dosed in Phase 2b clinical trial of ultevursen for USH2A-associated retinitis pigmentosa***

We are thrilled to share that [Sepul Bio](#), an innovative business unit of Laboratories Théa, will carry forward the groundbreaking work started by ProQR Therapeutics. [Today, Sepul Bio announced the first clinical participant has been dosed in the LUNA clinical study.](#) LUNA is a Phase 2b clinical study for ultevursen in individuals with Usher syndrome or non-syndromic RP due to variants in exon 13 of the Usher syndrome type 2a gene.

**LUNA**, also called SB-421a-006, is a two-year study testing **ultevursen**. Ultevursen is an experimental mRNA therapy aimed to help people with Usher syndrome type 2a maintain their vision. The LUNA study will enroll 81 participants, both adults and children over eight years old, who have retinitis pigmentosa caused by specific changes, or variants, in exon 13 of the *USH2A* gene. The first active clinical site is in the United States. Sepul Bio plans to open more sites in the coming months, sharing updates as they are confirmed.

#### **How does ultevursen work?**

Ultevursen is an RNA-based therapy that targets messenger RNA (mRNA). As covered in our [Science Education series](#), DNA is transcribed into RNA, which is then translated into proteins. mRNA carries the genetic instructions to make specific proteins. Stopping the translation process can prevent a protein from being made. This is what happens with variants in exon 13 of the *USH2A* gene.

Antisense oligonucleotides (AONs or ASOs) are short, custom pieces of DNA or RNA that bind to mRNA to block translation. Sepul Bio's ultevursen uses these custom RNA fragments to bind specifically to exon 13 of the *USH2A* gene. This prevents the mutated exon from being translated (a process called exon skipping), while the rest of the mRNA is processed normally. The result is a functional *USH2A* protein, even with the skipped mutation.

#### **Sepul Bio**

As a dedicated business unit of Théa, Sepul Bio is at the forefront of advancing transformative mRNA therapies for inherited retinal diseases, with a particular emphasis on the further development of two cutting-edge ophthalmic products—ultevursen and sepfarsen.

The Sepul Bio team was established in December 2023 and has worked hard to restart the ultevursen program. The clinical team looks forward to quickly enrolling the study, to advance ultevursen's development and bring this potential novel therapy to the USH2A patient community as quickly as possible.

Andrew Bolan, Patient Advocacy Director, is a key member of Sepul Bio who transitioned from ProQR. He continues to serve as a vital link in this work. Mr. Bolan will share updates with USH2A-affected individuals and families as significant progress is made. Interested individuals can send inquiries to [contact@sepulbio.com](mailto:contact@sepulbio.com).

This is also a good time to make sure you are a part of our [USH Trust](#) to be kept informed on this and other clinical trials. The USH Trust is our most powerful tool for supporting research. Make sure that you have updated your information in our [USH Trust](#). If you suspect your information is outdated, take a moment to update your preferred email address, your birthdate, your type and subtype, and whether you have genetic confirmation of this diagnosis.

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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**

On the "[Science News](#)," page you will find general news broken down by treatment type. We recommend you check the "[All Science News](#)" section to find short summaries using everyday language about all things Usher syndrome-related. Each summary contains a "What this means for Usher syndrome" section as well as a link to the original article.

A team of staff and volunteers work together to summarize research from highly respected, peer-reviewed medical journals in everyday language. Members of the Usher Syndrome Coalition Scientific Advisory Committee review the summaries for accuracy.

We hope that they are informative to members of the Usher syndrome community.

This work is ongoing; more study summaries will be added to this page over time. For questions or concerns please contact us at: [info@usher-syndrome.org](mailto:info@usher-syndrome.org)

*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.*

## ON WELL-BEING: Practicing Self-Care During the Holidays: Finding Balance Amid the Festivities

The holiday season is a time of joy and togetherness, but it can also bring stress and overwhelm. It's easy to put yourself last in the hustle of shopping, planning, and attending events. That's where self-care comes in. Taking time to care for yourself isn't selfish—it's essential, especially when managing the unique challenges of living with Usher syndrome.

### **What Is Self-Care?**

Self-care involves taking deliberate actions to care for your physical, emotional, and mental health. It's not about overhauling your routine or adding another item to your to-do list. Instead, it's about weaving small, meaningful practices into your day to recharge your mind and body. Studies show that people who practice regular self-care report lower levels of anxiety and higher satisfaction with life (Perrin et al., 2017).

### **The Science of Self-Care and Stress Relief**

Consider adopting the Danish concept of *hygge* (pronounced hoo-gah), which emphasizes coziness, comfort, and connection. Think warm blankets, soft lighting, and enjoying simple pleasures with loved ones. One study by Pressman et al. (2009) found that engaging in relaxing activities can lower cortisol levels—the stress hormone—and improve immune function. Whether it's taking a quiet walk, enjoying a cup of tea, or setting aside time for a favorite hobby, these small practices can have significant effects on mental health.

Incorporating *hygge* into your life can help slow things down and bring a little calm to the chaos. Research suggests that cultivating cozy, comforting environments can boost happiness and resilience—perfect for the holiday season.

## Tips for Holiday Self-Care

Here are some practical ways to nurture yourself this season:

- **Create a peaceful space:** Dedicate a corner of your home to relaxation—add soft textures, warm lighting, and calming scents.
- **Practice mindful breathing:** Spend a few minutes each day focusing on your breath to reduce stress and improve focus.
- **Set boundaries:** Say no to events or activities that feel overwhelming, and give yourself permission to rest.
- **Prioritize sleep:** Quality rest is crucial for recharging your body and mind. Consider creating a bedtime ritual to wind down.
- **Connect with others:** Lean on your support system and share meaningful moments with loved ones.
- **Self-Care and Resilience in the Usher Syndrome Community**

Practicing self-care is a powerful tool for building resilience. By addressing your own needs, you can better manage the challenges of the condition while staying grounded and hopeful. Whether it's joining a support group, carving out time for a personal hobby, or embracing hygge, self-care can provide a sense of control and peace.

As we navigate the holiday season, remember that taking care of yourself is not only beneficial for you—it also allows you to show up fully for those you care about.

You are not alone.

## References

- Perrin, P. B., et al. (2017). "The impact of self-care practices on stress and resilience." *Journal of Health Psychology*, 22(9), 1146-1155.
- Pressman, S. D., et al. (2009). "Relaxation and its impact on cortisol and immune function." *Journal of Psychosomatic Research*, 67(6), 573-581.

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We share the research and peer-reviewed literature that offers insight into well-being: the science behind staying grounded. Fill out this [poll](#) to request a topic.

**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 Life Hack of the Day

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

Looking for a last-minute holiday gift?! Wearing one of the [Usher syndrome definition t-shirts](#) is a low-effort way to spread awareness about Usher syndrome and educate the community.



### Our Contact Information

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