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USHER SYNDROME COALITION

CONNECTING THE GLOBAL USHER COMMUNITY

GROUNDED IN SCIENCE: July 2023

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

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A few reminders before getting to the good stuff. Let's dive in!

- Our USH Local Social in Boston has been rescheduled for Saturday, September 16th, opening up this gathering to more people in the community in honor of Usher Syndrome Awareness Day! Meet others with USH while trying out a variety of accessible activities taught by leaders living with Usher syndrome. Family members, partners, friends - all are welcome! Check out the locations for our other USH Local socials here.
- Speaking of Usher Syndrome Awareness Day, if you haven't already, please reach out to your governor to secure a proclamation for this day.
- A friendly reminder to join the Usher Syndrome Data Collection Program (USH DCP) because we need your help to expand and improve medical research. By generating the most comprehensive data collection effort for the Usher community, we can accelerate research and the development of treatments and therapies.
- 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

AAVantgarde Bio and Usher Syndrome Type 1B

Usher syndrome type 1B (USH1B) is caused by mutations in the Myosin VIIA (MYO7A) gene. This gene is large in size, which makes gene therapy challenging due to the limited packaging capacity of Adeno-Associated Viral (AAV) vectors. To overcome this challenge, researchers created a dual AAV8 vector platform to hold the MYO7A gene to be used as retinal gene therapy. These researchers have

been able to successfully demonstrate proof of concept using mice models as well as non-human primates. (Ferla et al.)

AAVantgarde Bio is an Italian-based international biotechnology company that uses Adeno-Associated Viral (AAV) vector platforms for gene delivery. A recent funding initiative was secured, allowing them to move forward with two of their programs, one of which is for USH1B, to establish a first-in-man proof of concept in patients with USH1B. (GlobeNewsWire).

For more, check out our Current USH Research page specific to USH subtype as well as gene-independent therapeutic approaches.

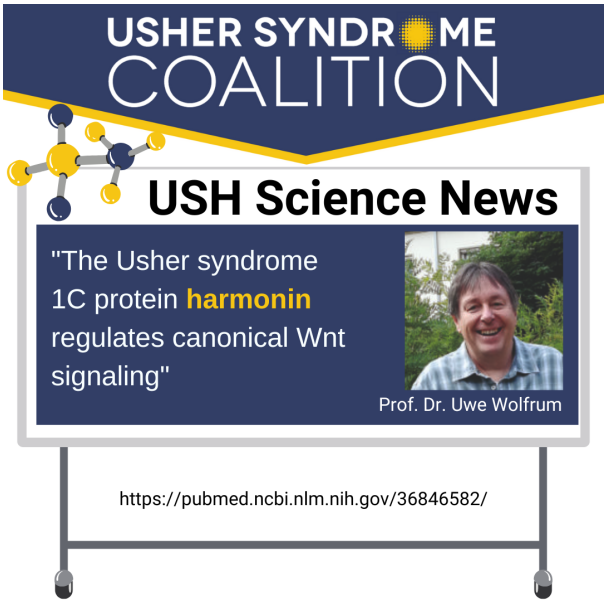
AAVANTGARDE BIO

View Current USH Research

IN CASE YOU MISSED IT: SCIENCE NEWS FEATURE

The Usher syndrome 1C protein harmonin regulates canonical Wnt signaling

The Wolfrum lab recently published a paper titled, "The Usher syndrome 1C protein harmonin regulates canonical Wnt signaling." We already know that the USH1C gene encodes the protein harmonin which organizes protein networks in the retina and inner ear. In the study, the researchers found that harmonin plays a crucial role in the canonical Wnt signaling pathway, which is important for maintaining the health and function of the eye and retina. The study found that without the harmonin protein functioning properly, this pathway is disrupted, which can lead to the development of retinal diseases and vision impairment.



What this means for Usher syndrome: This discovery is significant because it sheds light on the underlying mechanisms of Usher syndrome and could lead to the development of new treatments and therapies for this rare disorder. It also helps better understand the role Wnt signaling plays in the eye and retina and how it can contribute to retinal diseases.

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
Empowering Independence: Fostering Self-Determination

By Lanya McKittrick, Lane of Inquiry, Mom to 4 boys – Conner (24) and Dalton (15) have USH1B.

Parents, you may have wondered what the future might be like for your children. I know I did, especially early on, right after the diagnosis.

Questions you may have asked yourself might include: What kind of career will my child have? Will they find love? Will they have children, and what impact will the vision loss have on parenting? Will they be able to live independently?

Sometimes it can feel paralyzing to think about all this uncertainty. There is so much you can't control. But one thing you can do is to help your child be independent and to live a self-determined life.

Self-determination[1] plays a pivotal role in the lives of individuals with Usher syndrome. Self-determination refers to one's ability to make choices, set goals, and take control of their own lives. It enables them to take charge of their own destiny, make decisions aligned with their values, and actively participate in society. By fostering self-determination, you build resilience and help your child develop a sense of purpose. It promotes autonomy, enhances problem-solving skills, and boosts self-confidence.

Here are some strategies for parents in fostering self-determination:

- **Education and Awareness:** Teach your child about Usher syndrome and the available support systems so they can gain confidence in their abilities and make informed choices. Familiarize yourself with the Expanded Core Curriculum.
- **Self-Advocacy Skills:** Encourage your child to express their needs, assert their rights, and actively participate in decision-making processes related to their education, employment, and personal life.
- **Goal Setting and Planning:** Help your child to define their aspirations, whether it be pursuing education, career development, or engaging in recreational activities.
- **Assistive Technology (AT) and Accessibility:** Advancements such as screen readers, speech recognition software, and assistive devices enhance communication, access information, and facilitate independence. Make sure your child has the appropriate accommodations and AT.
- **Early Choice Making:** Find ways for young children to make simple choices such as what to wear each day – this helps make choice-making easier in later years.

As a parent of four boys, I know first-hand how difficult it can be to foster self-determination. Sometimes it's easier to make decisions yourself because you are simply trying to get through each day. But promoting self-determination at a young age can help both you and your child in the long run. By implementing strategies such as these, you can empower your children to assert their independence, make informed choices, and pursue their dreams.

[1] Self-determination is one of the 9 areas of the Expanded Core Curriculum, the "concepts and skills that often require specialized instruction with students who are blind or visually impaired in order to compensate for decreased opportunities to learn incidentally by observing others."

USH Life Hack of the Day

Send your USH life hacks to info@usher-syndrome.org

“Adding raised stickers (button-like) to my microwave on the '30 Second' and 'Timer' buttons are less stressful for my eyes as I just feel for the raised buttons.”

- Krista Webb

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