

Foundation Updates

2024 was a great year for the SHANK2 Foundation. The Schmidt Family organized a successful fundraiser over the summer, we hosted our annual SHANK2 Day conference in November, we posted a “newly diagnosed handbook,” and Samantha Lanjewar, PhD, joined the Board of Directors. 2025 is off to an equally great start with Michelle Murolo, PhD, joining the Board of Directors and taking on the role of liaison to COMBINEDBrain.

One goal this year is to increase our communication with the community on Foundation activities. We have had an amazing growth of individuals in our community, and while we still strive to know each and every individual personally, we are also working on broader methods of communication. Do not hesitate to reach out to us at any time: news@shank2.org



RARE-X Data Collection

In order for researchers and clinicians to study SHANK2, they need data! Specifically, they need basic information about the symptoms associated with various mutations.

One of the most important things you can do to help the SHANK2 Foundation is to enter your (or your child’s) information into the RARE-X data collection database.

The RARE-X platform is Institutional Review Board (IRB)-approved and is free of charge to patients and their families. Get started here:

<https://rare-x.org/shank2/>

SHANK2 Innovation Grant: Request for Proposals

This spring, the SHANK2 Foundation had its first request for research proposals. The SHANK2 Foundation’s \$25,000 Innovation Grant will support an early-stage study aimed at advancing novel research or therapeutic strategies for SHANK2-related disorders. Applications are due on April 7th, and the review of proposals will occur this spring, followed by an announcement of the awardee.



SHANK3 Gene Therapy News

A gene therapy trial was recently approved by the FDA for SHANK3 disorders. This study is being conducted by Jaguar, and we are actively following the outcome. The Foundation's President, Ben Hutz, PhD, recently spoke with their CEO. Here are some key takeaways:

- A similar gene therapy trial can theoretically be done in SHANK2.
- It is difficult to attract a company to attempt this type of therapy for two main reasons:
 - ❖ It is **very** expensive to run this type of trial.
 - ❖ It is difficult to measure the success of treating neurodevelopmental disorders.

A short-term goal of the SHANK2 Foundation is to make SHANK2 disorders as attractive as possible for a similar trial. The following activities will help accomplish this:

- ❖ Initiating a SHANK2 biobank
- ❖ Documenting the "disease burden" of families with a SHANK2 disorder
- ❖ Understanding the main symptoms of SHANK2 disorders and how best to measure them via the RARE-X Data Collection Program

Upcoming Activities



SHANK2 Biobank: Work has started on the first iPSC (induced pluripotent stem cell) SHANK2 line through the COMBINEDBrain Biorepository. We will start collecting blood and plasma samples for the biobank this summer, as well as begin a second iPSC line.

blinklab

The **blinklab study** on characterizing SHANK2 disorders via involuntary blink responses is recommencing and will be looking for additional participants.



A **community knowledge and medication survey** will soon to be sent out so that we can learn from each other's experiences.



Our **genetic counselor student intern**, Isabella Brown, from the jumpstart program will be posting community education materials on Facebook.

Educational Series!

Science



Parenting



Community



Week 1

Proteins

Facilitator: Michelle Murolo, PhD

Sunday, April 13th, 8:00pm ET

Zoom link:

<https://wesleyan.zoom.us/j/94713688372>

Week 2

Genomes

Facilitator: Michelle Murolo, PhD

Saturday, April 19th, 3:00pm ET

Zoom link:

<https://wesleyan.zoom.us/j/94713688372>

Week 3

SHANK2 Disorder Characteristics

Facilitator: Ben Hutz, PhD

Sunday, April 27, 9:00am ET

Zoom link:

<https://slu.zoom.us/j/95374501366>

Week 4

Stem Cells

Facilitator: Michelle Murolo, PhD

Sunday, May 4th, 8:00am ET

Zoom link:

<https://wesleyan.zoom.us/j/94713688372>

Look for additional information about each week's
Topic on the SHANK2 Foundation Facebook page!

