

## ***Fall 2025 Newsletter***

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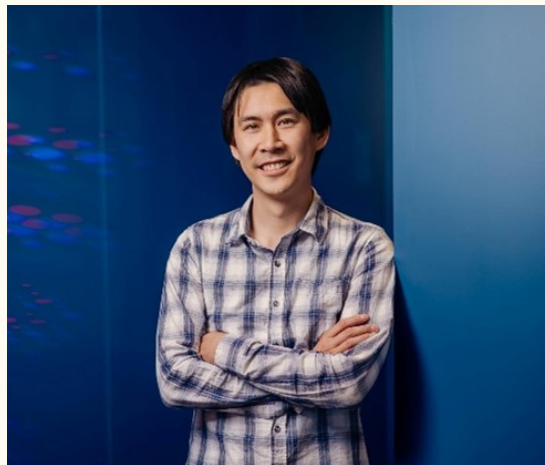
### **SHANK2 Day November 9, 2025**

Save the Date: Sunday, November 9th. We will be holding our annual fall SHANK2 Conference in early November this year. This conference brings together families, clinicians, and researchers to discuss SHANK2 disorders. It will be held virtually over Zoom and consist of a variety of activities ranging from informal introductions to formal research talks. Registration and participation is free!

[Register for SHANK2 Day](#)

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### **Announcing the 2025 Innovation Grant Recipient**



The Innovation Grant was created to support an early-stage research project aimed at advancing novel research or therapeutic strategies for SHANK2-related disorders. This award provides critical seed funding for researchers to turn their visionary idea into an impactful project. The recipient of this year's award is Dr. Aaron Kuan, Assistant Professor of Neuroscience and Biomedical Engineering at Yale University.

## **Project Title: A Novel Pan-Expansion Microscopy Approach to Bridge Molecular Alterations and Circuit-level Dysfunction Underlying SHANK2-Related Disorders**

*SHANK2* is a gene that regulates the production of the SHANK2 protein, which plays a key role in how brain cells (neurons) connect and communicate with each other. When there is a change (mutation) in *SHANK2*, it can cause disruptions that lead to intellectual disability, autism spectrum disorder, communication delays, and other neurodevelopmental conditions.

Dr. Kuan's research project aims to better understand how mutations in *SHANK2* affect the brain, including at the synapses, which are tiny structures where neurons pass signals between each other. When these connections don't form or function properly, the brain's ability to process information can be impaired, affecting learning, memory, and behavior.

To study this, Dr. Kuan's team is using a new and advanced imaging technique called pan-Expansion Microscopy (pan-ExM). This cutting-edge method allows for a zoomed in visualization of incredibly small structures in the brain that were previously too small to study in detail. With this powerful tool, the effect of *SHANK2* mutations in the brain can be determined at a molecular level.

Ultimately, the goal of this project is to create detailed maps that show how *SHANK2* mutations change brain structure and function. These insights could lead to the discovery of new biomarkers and therapeutic targets to better diagnose and treat SHANK2-related disorders. By understanding what is structurally different in the brain due to these mutations, researchers can begin to explore strategies to overcome these differences.

For questions about the innovation award, please contact Alex Lanjewar at [alex@shank2.org](mailto:alex@shank2.org)

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### **Building a SHANK2 Biorepository**

Over the past several months, The SHANK2 Foundation has been working to establish a biorepository of blood samples from individuals with SHANK2 variants. A biorepository is simply a professionally managed collection of biological samples stored for scientific research. In our case, the samples are housed at the Van Andel Institute through a partnership with COMBINEDBrain.

COMBINEDBrain is a consortium of organizations focused on rare genetic neurological disorders. By pooling efforts and resources, COMBINEDBrain makes it possible for smaller groups like ours to participate in projects and research initiative. COMBINEDBrain has been holding blood draw collections in select cities across the United States.

### **Why is establishing a biorepository important?**

Researchers need samples to analyze! By comparing blood from individuals with SHANK2 variants to samples from neurotypical individuals, scientists can learn more about the processes that are altered in those with SHANK2 variants.

If we can reach 20 samples in our SHANK2 biorepository, we will qualify to take part in a COMBINEDBrain organized proteomics biomarker project planned for spring 2026. This study will involve 10–15 pharmaceutical and biotech companies who will be analyzing the biorepository samples.

### **What is a proteomics biomarker study?**

Proteomics is the study of proteins, essential molecules that keep our cells functioning and do a lot of the work in our cells. SHANK2 itself is a protein found in neurons. In a proteomics biomarker study, researchers compare protein profiles between SHANK2 and neurotypical individuals to identify biomarkers—measurable indicators of a biological condition.

Discovering a distinct protein signature for SHANK2 could provide an objective tool for tracking treatment response. Having an objective measure is very important for companies working on drug development, drug repurposing, and genetic therapies.

### **How are samples protected?**

A Biorepository Governing Committee reviews all sample requests and controls sample distribution. The Committee is composed of two COMBINEDBrain staff members, two COMBINEDBrain patient advocacy group member leaders, and (for SHANK2 related samples) a pre-designated representative from The SHANK2 Foundation. All members of the Committee have had human subjects clinical research training and must unanimously agree to approve a sample request.

### **Where are we now and what's next?**

Currently, the SHANK2 biorepository contains samples from seven individuals. We are deeply grateful to these families for their participation! However, we still need 13 more samples to reach the 20 required for the proteomics study.

Unfortunately, families cannot simply donate blood at a local lab like Quest or LabCorp because there is no physician's requisition order for the blood draw. Samples must be collected through COMBINEDBrain organized blood draws. While remote home blood draws are possible, they are very costly and not in our current budget.

That's why we need your help: if a COMBINEDBrain blood draw takes place in a city near you, please consider participating. Each contribution brings us closer to advancing SHANK2 research and opening doors to future treatment possibilities.

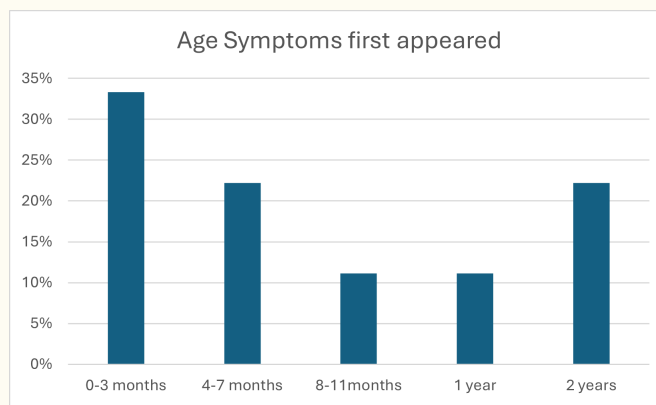
For questions about the biorepository, please contact Michelle Murolo at [michelle@shank2.org](mailto:michelle@shank2.org)

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## **SHANK2 Data Collection Program through RARE-X**

We're excited to share an update on our data collection program run through RARE-X (Global Genes). First a quick overview. The SHANK2 Data Collection Program is a patient registry containing information on SHANK2 individuals including, basic demographics, genetics, and a variety of research validated surveys. This program is run through RARE-X, which is part of Global Genes, a non-profit organization dedicated to supporting rare diseases. The program is overseen by an Institution Review Board and held to the strictest standards of patient privacy. All individuals maintain complete control over their information and only de-identified information can ever be shared with researchers. The information provided in this patient registry is the single most important thing families can contribute to pushing forward research on SHANK2 disorders. This data can be used for everything from forming a basic clinical picture of what it means to have (or diagnose) a SHANK2 disorder, to helping generate metrics that can be used in clinical trials.

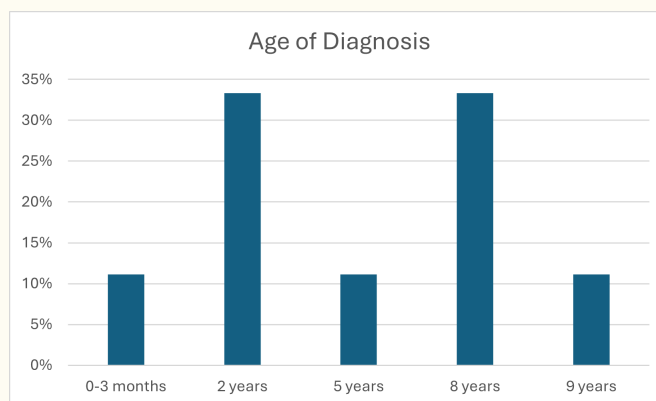
The SHANK2 Data Collection Program was launched in July of 2024, replacing our previous patient registry. There are currently 32 individuals registered from 9 different countries. First, we look at diagnosing SHANK2 disorders. The following chart summarizes the age symptoms were first noticed.



The next chart gives the age when a SHANK2 diagnosis was given. Note that while symptoms are generally noticed very early, it can take years before a formal diagnosis is given. It would be great if we could narrow this gap and improve the diagnoses of SHANK2 disorders. The first symptoms noticed were typically one of the following:

- Communication
- Fine/Gross Motor
- Eye Tracking
- Over or Under Reactive
- Repetitive Behaviors
- Delayed Babbling

However, this next chart shows that the age of diagnosis is typically significantly later. It would be great if we could narrow this gap and improve the diagnosis of SHANK2 disorders.



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

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. Only de-identified information is shared with researchers and you always have control of your data.

The RARE-X platform is Institutional Review Board (IRB)-approved and is free to patients and their families.

[Get Started here](#)

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## **Community Survey: Our Collective Knowledge**

Welcome! As our community approaches 300 members, we've collectively amassed a wealth of experience and insights regarding SHANK2 disorders. This shared knowledge, born from our individual journeys in supporting our children and ourselves, holds immense potential. To harness this power and advance our understanding, we've created a survey. Your participation will contribute directly to identifying key research areas, uncovering potential mechanisms, and exploring promising treatment avenues. By sharing your experiences, you'll help shape the future of SHANK2 research and empower our community to better support those affected.

The survey is anonymous unless you choose to share your contact information with us.

[Take the survey](#)

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## **Schmidt Golf Outing Fundraiser**

We are grateful for the Schmidts (Sarah and Pete) who ran their second annual golf outing fundraiser in August. With their incredible support, we have been able to offer our first ever SHANK2 research grant as well as continue our efforts in the SHANK2 biorepository. If anyone is interested in running their own fundraisers, Sarah and Pete would be happy to talk with you and help you get started.

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## **SHANK2 Phenotyping Study Published**

In 2021, the SHANK2 Foundation began collaborating with the Sever Autism Center at Mt. Sinai Medical Center. Through the generous donation of Polly Appel and her husband Peter, we were able to fund a 10-person study to investigate the clinical presentation of SHANK2 disorders (phenotype). The SHANK2 community came together and 10 participants completed the study. We're pleased to announce that the final results of that study have been published in the Journal of Neurodevelopmental Disorders and is publicly available.

[Ready the Summary](#)

[Read the Full Article](#)

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### **SHANK2 Educational Series**

In the spring, we had a 4-week educational series that brought together families to learn more about the science related to SHANK2 disorders and to talk about parenting kids with SHANK2 variants. Michelle Murolo created educational videos on Proteins, Genomes, and Stem Cells. You can access the videos on the Foundation's Vimeo page. Most importantly, it was a safe space where we could talk about the struggles in raising kids with SHANK2 variants, offer suggestions and tips to each other, and support each other. Based on feedback from these sessions, we're thinking about starting a monthly pop-in support session via zoom.

[Access the recorded videos](#)

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**The SHANK2 Foundation Facebook group**

We have an active Facebook private group named, “The SHANK2 Foundation”. If you’re not already a member, join us to ask questions, share tips, and build community with other SHANK2 families!

[Join the Group](#)

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**The SHANK2 Foundation**

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