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Help raise awareness, donate today

February is a RARE month!

We hope you are enjoying our social media countdown to International SCN2A Awareness Day on February 24th! We are thrilled to be partnering with passionate advocates from around the globe to share countdown photos of international SCN2A warriors as well as SCN2A Facts of the Day.

We invite you to educate yourself and others about this rare condition through the **SCN2A Fact of the Day** initiative. Knowledge is power, and by sharing these insights, we can create a more informed and supportive community.

Stay tuned for exciting updates and major announcements by following us closely on social media or [sign up here for email and text alerts](#). You won't want to miss what's coming next!

Let's come together to make a lasting impact. Read, learn, and share—because awareness is the first step towards understanding, change, and cures!

Two Grants open on SCN2A Awareness Day, 2/24/2024



Visit our website:
www.scn2a.org/professionals.html
for more details.

2024 Action Potential Grant Cycle

- ▶ Awarding two (2) \$75,000 grants
- ▶ Early career scientists/clinical researchers
- ▶ Supporting research that advances understanding of the cellular, molecular, genetic, and systems-level mechanisms of SCN2A-related disorders.
- ▶ Grant cycle opens on International SCN2A Awareness Day, February 24th



 IN RECENT NEWS

Congratulations to Paul Jenkins, PhD at the University of Michigan, Winner of this Year's Million Dollar Bike Ride Pilot Grant Award for SCN2A for his outstanding proposal "Investigating Channel Scaffolding and its Contribution to SCN2A Disorders." Paul was awarded the \$61,280 raised during the 2023 MDBR by the team pictured below.

In his groundbreaking research, Paul plans to unravel the mysteries behind SCN2A variants that might not exhibit detectable effects on channel functional properties but are still pathogenic. His proposal delves into the hypothesis that these variants contribute to disease phenotypes by disrupting normal channel scaffolding, leading to channel mislocation and neuronal dysfunction. This innovative approach has the potential to discover new insights into SCN2A-Related Disorders (SRD), paving the way for advancements in our understanding of the condition and potentially new targets for therapeutics.

The 2024 MDBR is June 8th!

Save the date to ride or volunteer with the SCN2A Warrior Riders in Philadelphia! It's a fun event for all ages with local restaurants providing amazing food. Can't travel? Set up a virtual ride with your friends. Details and registration coming soon.



Top autism-linked genes join forces to shape synaptic plasticity | [The Transmitter: Neuroscience News and Perspectives](#) This January 29th article by Holly Barker provides a helpful overview of the recent study published by Paul Jenkins, Kevin Bender, Stephan Sanders, et al, in the journal *Neuron*.

"It's a keystone gene, so if we pull on the SCN2A thread, we might understand more about the actual mechanisms that underlie ASD." - Kevin Bender, PhD, UCSF

So far, 12 states in the US have proclaimed February 24th as SCN2A Awareness Day!

Breaking news...Texas and Georgia are in! Final updates coming 2/24.

Will your state be on the list this year?

For information about how to advocate for SCN2A in your state, [send us an email](#).



 **BEYOND THE DIAGNOSIS**



RARE DISEASE CURES
ACCELERATOR
**DATA AND ANALYTICS
PLATFORM**



What happens to your data?

The FamilieSCN2A Foundation is excited to announce a significant step towards enhancing research efforts for SCN2A-related disorders: A data-sharing agreement that will integrate vital data from the SCN2A Clinical Trial Readiness Study (CTRS) into the Rare Disease Cures Accelerator-Data and Analytics Platform (RDCA-DAP®). This collaboration aligns with our mission to improve the lives of those affected by SRDs by accelerating research. We believe that sharing data is the fastest way to get the most effective treatments to the most people. As our CSO, Shawn Egan, says, "This gets us more shots on goal!"

[RDCA-DAP®](#) is an FDA-funded initiative that provides a centralized and standardized infrastructure to support and accelerate rare disease characterization, with the goal of accelerating therapy development across rare diseases. In partnership with the National Organization for Rare Disorders ([NORD](#)) and The Critical Path Institute ([C-Path](#)), RDCA-DAP® promotes the sharing of existing patient-level data and encourages standardization of new data collection.

We spoke with Heidi Grabenstatter, PhD, Science Director for the Data Analytics Platform. Heidi's role is to oversee the onboarding of data for several rare disorders, including SCN2A. As a neutral convener of public/private partnerships, the RDCA-DAP aims to identify and improve bottlenecks in drug development by creating tools across diseases to aggregate and analyze data. More data plus better tools for analysis equals faster discoveries that can lead to cures.

"We are excited to have the SCN2A CTRS data and grateful for the patients and their families who took the time to participate in the data collection efforts. We recognize how hard that is," says Heidi, who also served as the Science Director for the International Foundation for CDKL5 Research and is a person with epilepsy.

Heidi shares that often, data may be available (from academic research, clinical trials, natural history studies, etc.), but until it is pulled together in a useful format, it's not always helpful. By integrating such data in a regulatory-grade format suitable for analytics, RDCA-DAP accelerates the understanding of disease progression, clinical outcome measures, and biomarkers, and facilitates the development of mathematical models of disease and innovative clinical trial designs.

We support the RDCA-DAP's thinking that there is an incentive for all groups involved in data collection to share data. Heidi explains, "RDCA-DAP is looking into how we could provide additional help in the rare epilepsy space—there is a need for data standardization and harmonization across available data sources to ensure it is fit-for-purpose for drug development and to inform future data collection efforts. C-Path has a long-standing experience collaborating with industry and FDA."

C-Path is a global organization with a mission to lead collaborations that advance better treatments for people worldwide. We know that SCN2A does not discriminate based on where you live—55 countries that we know of, so far.

So much data exists in silos, imagine the power it will have when it's collected in one place, accessible to those who can use it to accelerate treatments. The FamilieSCN2A Foundation believes that all data* should be accessible to the patients who provide it and to those working towards cures. This agreement with RDCA-DAP is a first step in that direction. We appeal to ALL researchers and industry groups holding SCN2A data to share it with RDCA-DAP.

Please contact hgrabenstatter@c-path.org to learn more.

**All shared data is de-identified before it is released to RDCA-DAP. Do you have the [CRID \(Clinical Research ID\)](#) yet?*



The banner features a background image of a man in a white shirt and backpack at a booth. Text on the banner includes: 'FAMILIESCN2A FOUNDATION WWW.SCN2A.ORG', 'SCN2A Town Hall Meetings', 'CONNECT | SUPPORT | RESEARCH', 'Our Goals', 'Ask me: What's ERG...', and 'Meeting Leader: Shawn Egan, PhD'. A purple wave graphic at the bottom contains a white icon of three people and the text 'Open forum to ask questions about research and or community resources.'

What is the SCN2A Town Hall all about?

It's a private Zoom call (not recorded) with FamilieSCN2A leaders for families affected by SCN2A-related disorders. Designed to share information, challenges, and ideas about all things SRD, the calls are held bi-monthly on the 5th and 20th at differing times to make them accessible to a variety of time zones and schedules. Often there will be a topic, such as clinical trial design, but the goal is to encourage community dialog. Bring your questions and experiences!

Families: email info@scn2a.org for a link to register.

Shop quickly for your International SCN2A Awareness Day and State-specific Proclamation t-shirts!

The SCN2A store is always open



Help raise awareness, donate today



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