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Another conference with record-breaking registration!!

CHARTING THE COURSE
WWW.SCN2A.ORG

INTERNATIONAL SCN2A
FAMILY & PROFESSIONAL CONFERENCE

JULY 19-22, 2023
SHERATON BOSTON HOTEL 39 DALTON STREET, BOSTON, MA

Brought to you by:
 FAMILIESCN2A
SCN2A-RELATED AUTISM & EPILEPSY

Co-hosted by:
 ROSAMUND
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Virtual Registration (closes July 16th @11:59 ET)

Detailed Agenda



RESEARCH opportunities at the conference in Boston! [Click here to learn more and reserve your spot.](#)

Participation in research is the single most important thing (after fundraising) you can do to help chart the course towards a cure. We are grateful for those who consider getting involved, whether it's filling out a survey or donning an EEG helmet. Your efforts positively affect the entire FamilieSCN2A community! *If you're attending the conference in person, please check your email for updates and other opportunities to participate.*

WHAT'S HAPPENING?



We are excited to announce the **3rd Annual Golf Fore SCN2A Tournament** to raise money for the FamilieSCN2A Foundation. Please join us on Monday, August 21st at Springville Country Club in Springville, NY.

SCN2A Families: Please come even if you don't golf! It's a fun place to socialize with other families and the golfers love to meet the people they are supporting!

We are grateful to Golf Committee members and the Egan family, whose 6-year-old daughter, Harper, has an SCN2A-Related Disorder (SRD), for hosting this wonderful event! It's the passionate commitment of families and their circles of friends, colleagues, and companies, that will make the difference in funding research to cure all SRDs while improving the lives of all who

have been affected.

Can't make it? Your tax-deductible donation to the organization is greatly appreciated.

BEYOND SEIZURES, WHAT REALLY MATTERS?

THE INCHSTONE PROJECT

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LEND YOUR VOICE!
Help identify non-seizure outcomes of importance to DEE families

Go to screen.inchstoneproject.org and Use Access Code **BAND** to See if This Survey is Right For You!

Our friends at [The Inchstone Project](#) invite you to participate in a research study specifically focused on meeting the needs of children and young people with severe to profound impairment.

The criteria for participating:

Must be at least 1 year old **AND** have very severe communication challenges (largely or completely nonverbal and cannot communicate effectively even nonverbally).

AND have either or both of the following:

Severe challenges in mobility (require a stroller or to be carried, even at home) and/or dependence on a feeding tube for nutrition or on someone to feed them.

[Click here](#) and enter code **BAND** to see if you qualify. Gift cards are available upon completion.



What will *your* team do? Registration opens July 16th

Join us (virtually) on or around October 21-22 for the 4th [Annual SCN2A Warrior Challenge](#). You can run, walk, roll or stroll to support the FamilieSCN2A Foundation!

This is a virtual event, so design the course however you want to challenge yourself and your friends: Is it 100 meters, a mile, a 5K; are you looking for a good reason to take on a 10K or something longer? Whatever the distance and location, join families from all over as we run, walk, roll in a wheelchair or push a stroller to complete the SCN2A Warrior Challenge for a great cause!



Clinical Trials

We are excited by the prospects for clinical trials in the pipeline for SRDs! Right now there are three trials recruiting for patients: Praxis 222 Embrave, Praxis 562 Embold (both specific to SCN2A gain of function), and Longboard Pharmaceuticals, The Pacific Study (for DEEs, not SCN2A specific). [CLICK HERE TO LEARN MORE](#)



IN RECENT NEWS



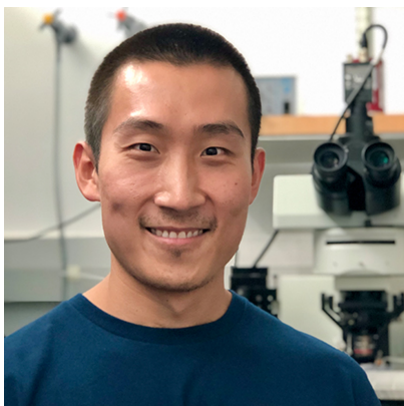
Hodgkin-Huxley Research Grant

DID YOU SEE THE [PRESS RELEASE](#)?!

Congratulations to [Yang Yang, PhD](#), of Purdue University, recipient of the FamilieSCN2A Foundation's Inaugural Hodgkin-Huxley Research award in the amount of **\$2.5 Million**. The five-year grant will support Dr. Yang's research aimed at developing personalized treatments for a splice site SCN2A variant, pushing forward our understanding of this understudied area of SRDs. The grant was fully funded through a generous donation made by an individual family.



Congratulations to Xiaoling Chen, PhD of Purdue University, for becoming an [American Epilepsy Society 2023 Early Career Fellowship Awardee](#)! The FamilieSCN2A Foundation and AES are proud to announce the co-funding of this Postdoctoral Research Fellowship in the amount of \$50,000. Dr. Chen will be evaluating the anti-seizure efficacy of AAV-KCN1A in Scn2a-deficient mice. We are excited to continue our support of this past [Action Potential Grant Awardee](#). Dr. Chen's mentor, Yang Yang, PhD, is also dedicated to SRD research (see above!).



The Foundation's [Action Potential Grant](#) has funded several brilliant, early career scientists since its inception in 2018. In this published preprint, 2022 AP Grant recipient, Chenyu Wang, and colleagues, showcase their research into Vestibulo-ocular reflex (VOR) in Loss of Function SCN2A-related disorders. This is critical to not only understanding which cells and synapses are affected in SCN2A but may potentially be used as a biomarker in clinical trials! Attendees of last year's FamilieSCN2A Summer Seminar in Ohio participated in some of this research and it will continue at this year's conference in Boston. Congratulations to the UCSF Bender lab, Chenyu and Guy Bouvier who will have his own lab in Paris!

[Impaired cerebellar plasticity hypersensitizes sensory reflexes in SCN2A-associated ASD | bioRxiv](#)

Another Recent SCN2A Publication

Al George, Dennis Lal, Jen Pan, et al. ["Scanning mutagenesis of the voltage-gated sodium channel NaV1.2 using base editing"](#)

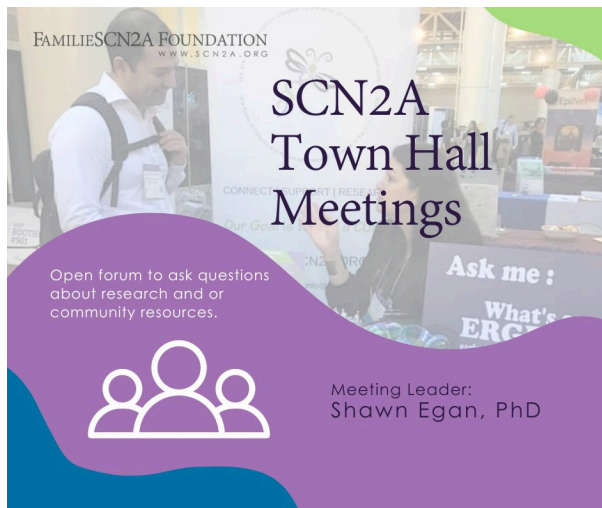
 MONTHLY RECAP



The Million Dollar Bike Ride was a HUGE success, raising \$60K for FamilieSCN2A research

The wildfire smoke blew away just in time for a beautiful ride through Philadelphia on June 10. An impressive turnout of 10- and 30-mile SCN2A Warrior Riders plus a slew of volunteers and cheerleaders rallied to push us over the finish line. We met UPenn's Orphan Disease Center's \$30K matching goal and can't wait to see who gets this year's \$60K SCN2A MDBR grant.

THANK YOU! PLEASE JOIN US NEXT YEAR!



SCN2A TOWN HALL DATES LED BY SHAWN EGAN, PHD

Register once in advance for all meetings:

Jul 15, 2023 11:00 AM

Aug 18, 2023 08:00 PM

Sep 15, 2023 06:00 PM

Oct 15, 2023 12:00 PM

Nov 15, 2023 06:00 PM

Times are Eastern Standard. After registering, you will receive a confirmation email with the link to join. These sessions are for **families only** and are not recorded.



Sodium Channel Advocates Take Denmark

The 2nd *European SCN8A & SCN2A Conference and Family Gathering* in May was a great success! SCN2A families representing 15 countries attended and strong, collaborative relationships were renewed. The research community heard loud and clear that we are "Stronger Together, Against All Odds," especially when patient advocates are included in the process. The FamilieSCN2A Foundation was pleased to encourage attendance by providing travel scholarships. Special thanks to the family organizers from [SCN2A Italia](#) and [SCN2A Germany](#), Stefania Dantone and Svenja Kaden!



BEYOND THE DIAGNOSIS



Chances are you have seen Amanda Gale's volunteer work all over social media. The warrior mom behind the Warrior Wednesday feature, Amanda loves interacting with SRD families and hearing

their stories of struggle and triumph. "Our worlds can be heavy," she says, "So I appreciate being able to add a little sunshine to every week." She especially enjoys reaching out to newer families in the community and shining a positive light on their kids.

As a Special Educator working with kids, Amanda knew there was something different about her son Henry's autism. While doctors brushed aside her concerns, she pushed for more testing and finally got the SCN2A diagnosis in 2019 when Henry was 2. The neurologist basically said, "good luck," so she reached out to the FamilieSCN2A Foundation for help in understanding what it means to have an SRD. "It opened up a whole community I never knew I would have!"

"People need to step up!"

When she realized the organization was parent-driven, Amanda thought she could help by putting her passion for community outreach to work. She recalled someone from Foundation saying, "When you're a special needs parent you feel helpless, but when you give back it makes you feel like you can take on the world! You find the power to keep that momentum pushing you forward." She adds, "On days when I'm doing things for FamilieSCN2A or I'm at the Maine State House advocating for higher pay for special ed teachers, those are the days I feel my world isn't crumbling. I'm not thinking about 10 years from now but today and tomorrow and what makes a difference to people right now."

"Change needs to happen now because the world isn't ready for our kids."

At the conference in Boston, look for Amanda on the dance floor where she's organizing Friday night's party and celebrating as a member of the event planning task force. Fun fact about Amanda: She grew up in Australia in a small coastal town not unlike where she now lives in Maine with her husband and three kids. Next school year she's excited to have Henry in kindergarten down the hall from her!

Thank you, Amanda, for all you do!

If you would like to have your SCN2A Warrior featured one Wednesday and/or add your story to our collection, please email info@scn2a.org. Sharing our stories brings us together, raises awareness and moves the mission forward. We can't wait to hear yours.

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