SCN2A-RELATED AUTISM & EPILEPSY

FAMILIESCN2A®

FOUNDATION

WWW.SCN2A.ORG

WHAT'S HAPPENING

SCN2A Warrior Riders Registration and Donation Page

2024 Million Dollar Bike Ride

June 8th in Philadelphia

We're reaching out to you, our incredible community, to help build our team! We need virtual and in-person cyclists, in-person volunteers, and donations to make this event a success. Your involvement will not only raise awareness but also provide vital funds to drive research forward. **Each dollar our team raises will be doubled (up to $30,000)** by the **ODC,** turning $30,000 dollars into $60,000 dollars to fund critical research to improve the lives of those living with SCN2A-related disorders!

Help Spread the Word! Use this email template to encourage your family and friends to support the SCN2A community in this incredible event!

2024 International
SCN2A Family & Professional Conference

July 31 - August 3 — FAMILIESCN2A FOUNDATION 2024 SUMMER CONFERENCE — Anaheim, California

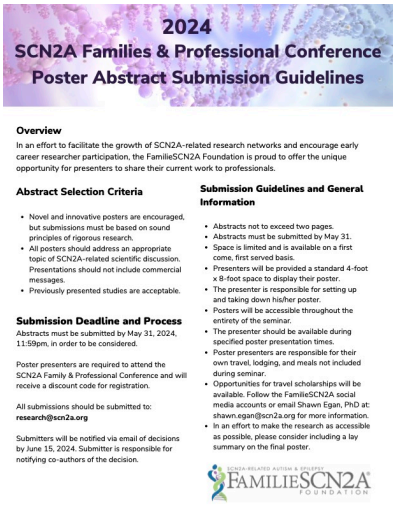
Wednesday 7/31:	8-5p Professionals Research Roundtable 2-5p Family arrivals / registration
Thursday 8/1:	8-4p General Session 8-4p Super Sibling Camp 6-9p Family Impact Dinner
Friday 8/2:	8-4p General Session 8-4p Super Sibling Camp 6-9p Warrior Dance Party & Dinner
Saturday 8/3:	Optional Disneyland Day
Sunday 8/4:	Departures

Join us at the 2024 SCN2A Family & Professional Conference

Happening from July 31st - August 3rd at Sheraton Park Hotel in the Anaheim Resort! This annual conference is a unique opportunity to unite the scientific, medical, and patient communities to advance our shared goal of finding better treatments and cures for SCN2A-related disorders. From informative presentations to interactive breakout sessions and networking opportunities, this event is packed with valuable insights, resources and special activities.

Connect, learn, and have fun! Register by May 31st for an unforgettable experience! **Rooms will sell out**, book your hotel room HERE, today.

Conference Registration



Submit your Poster Abstract

Researchers, showcase your work at the 2024 SCN2A Families & Professional Conference. Submit your poster abstract **by May 31st.** This is a fantastic opportunity to share your findings, connect with fellow experts and stakeholders and contribute to the advancement of SCN2A research.

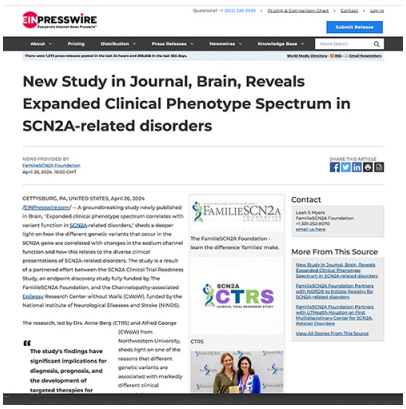
Poster Abstract Guidelines

Meet the Newest Addition to the FamilieSCN2A Foundation Team

Please join us in welcoming Irie Long to the FamilieSCN2A Foundation's team in the role of Program Manager.

Get to know Irie HERE!

SCN2A IN THE NEWS



Family participation in the SCN2A Clinical Trial Readiness Study continues to drive research forward on the relationship between gene function and clinical presentation of patients. Study published in *Brain* on April 23, 2024!

A recent publication in the journal *Brain*, titled "Expanded Clinical Phenotype Spectrum Correlates with Variant Function in SCN2A-related Disorders," is the culmination of years of collaborative work involving patients, caregivers, researchers, clinicians, and The FamilieSCN2A Foundation.

This work was achieved through a strategic partnership between the SCN2A Clinical Trial Readiness Study (CTRS), funded by The FamilieSCN2A Foundation, and the Channelopathy-associated Epilepsy Research Center without Walls (CWoW), funded by the National Institute of Neurological Diseases and Stroke (NINDS).

The CTRS enrolled 81 SCN2A patients worldwide through The FamilieSCN2A Foundation and meticulously collected detailed patient clinical information, including their specific SCN2A variant. Subsequently, sixty-nine unique SCN2A variants from the study were systematically examined through the CWoW to characterize their functional status. By combining clinical data with SCN2A functional status, researchers uncovered novel insights into SCN2A while validating many pre-existing understandings of the condition.

Key Study Findings:

- The study reaffirms that specific alterations in the SCN2A gene correspond to distinct clinical manifestations in individuals with SCN2A-related disorders. The primary phenotype strongly correlates with variant function, demonstrating statistical significance ($p<0.0001$).
- A spectrum of effects of SCN2A variants on sodium channel function was observed, ranging from hyperactive channels to completely inactive channels.
- Importantly, the clinical presentation of affected children varied based on the functional impact on the sodium channel. Hyperactive channels were typically associated with seizure onset in the first week of life, while more impaired channel function correlated with later seizure onset. Notably, almost all individuals without seizures had completely inactive sodium channels.
- Epileptic spasms were significantly more prevalent in infant-onset compared to neonatal or later-onset epilepsy ($p=0.007$).
- All children with SCN2A exhibited severe impairment, with even those with the mildest severity indices scoring significantly below the norm-referenced mean on the Vineland adaptive behavior composite score (average score of 49.5, >3 standard deviations below the test's norm-referenced mean).
- Non-seizure severity was highest in the neonatal-onset group and lowest in the autism group ($p=0.002$). As the age at seizure onset increased and channels became less active, the severity of neurological impairments tended to decrease.

This study and its findings hold implications for prognosis, clinical care, medical management, and drug development for SCN2A-related disorders. Providing caregivers and clinicians with a comprehensive understanding of the clinical and variant characterization of this disease equips them with valuable tools for managing affected individuals. Moreover, these insights can inform optimal trial design for SCN2A-related disorders, ensuring that appropriate drugs are tested in the right patient populations. Additionally, understanding the severity and natural history of the condition in relation to SCN2A variant function can underscore the urgency for regulators such as the FDA and EMA to thoughtfully consider clinical studies for SCN2A-related disorders.

The FamilieSCN2A Foundation is incredibly grateful to every single person involved in this important study. Research like this is critical to fulfilling our vision of a world with effective treatments and cures for all SCN2A-related disorders. It doesn't stop here, the next step in advancing our mission is The Dragonfly Study.

SCN2A-focused care at UTHealth Houston

The world's first center where families can quickly get up to speed on how to manage their child's condition is here! UTHealth Houston, led by faculty members from McGovern Medical School, caters to both children and adults with SCN2A-related disorders. Specialized care includes genetic counseling, tailored treatments for epilepsy, autism, and movement disorders, along with access to other subspecialties that are pertinent to that individual. The center operates at UT Physicians Pediatric Specialists - Texas Medical Center. Learn More HERE!

GET INVOLVED

Dragonfly Study is Launching Soon

The Dragonfly Study is an international patient-centered registry for SCN2A-related disorders crafted by researchers, medical experts, and caregivers. It is designed to capture the most relevant information essential for advancing research and sharing the impactful narratives of our community. Your involvement in the Dragonfly Study will play a vital role in shaping the future of SCN2A-related disorders research.
Learn more about the Dragonfly Study HERE!

Calling all Super Siblings!

Join our Super Sibling Mailing List and stay in the know...

Super Sibling Mailing List Form

Volunteer with FamilieSCN2A Foundation

Volunteers play a vital role in our organization by helping us fulfill our mission! We are currently seeking volunteers to assist with various initiatives including fundraising events, community outreach, social media campaigns, and more. To express interest in volunteering or learn more about available opportunities, please email our Program Manager, Irie Long.

COMING UP

Charity Ride in Honor of FamileSCN2A

Calling all NY friends! Join us at CycleBar Huntington on Saturday, June 1st at 12:30 pm for a charity ride benefiting FamilieSCN2A Foundation. Let's come together as a community to support families affected by SCN2A-related disorders. Sign up now and let's pedal for a brighter future!

Sign Up

4th Annual Golf Fore SCN2A Event

Mark your calendars! The **4th Annual Golf Fore SCN2A Tournament** is coming up on August 19th, 2024 at Springville Country Club, NY. Join us on the green as we raise funds for vital research and support for SCN2A-related disorders. Save the Date and stay tuned for more details!

If you cannot attend the event, please consider a tax-deductible donation to our organization. Your support is appreciated. **Employer matched donations here.**

Donate

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