The 2023 Annual Campaign is in full swing and we are 1/3 of the way to our $80,000 goal for our 8th year as a rare disease nonprofit!

As we chart the course in 2023, there is a tremendous need to have recurring donors on board to sustain the operation of our mission to accelerate research, build community, and advocate to improve the lives of those affected by SCN2A-related disorders (SRD).

By donating today and checking the box to make it monthly, you have the power to make our vision of a world with effective treatments and cures for ALL SRDs a reality FASTER, before another child is lost.

APRIL IS AUTISM AWARENESS MONTH

Throughout the month check social media and our website for informational posts about Autism-related SRDs. We encourage you to take 5 minutes to become more informed and then give 5 **or more** dollars toward providing valuable resources to the SRD community.

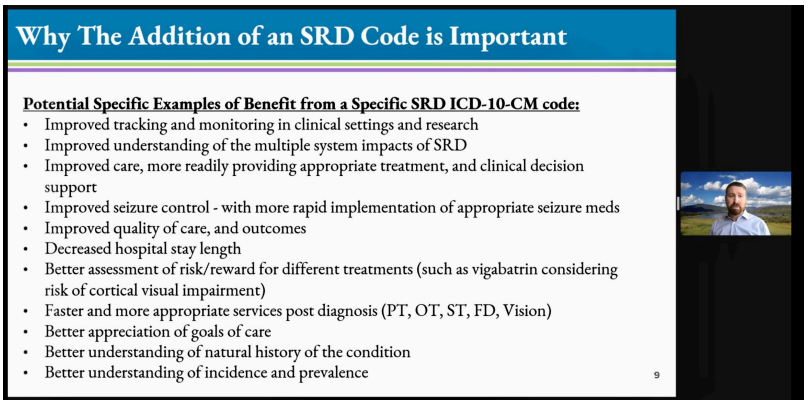
ICYMI: Watch and share our short documentary *Why "Families" is part of our name: SCN2A-related disorders affect the entire family*

DONATE

WHAT'S HAPPENING?

Register Today!

SCN2A-related disorders (SRD) is a global problem that demands global solutions. In an effort to make our 2023 SCN2A Family & Professional Conference as accessible, inclusive and diverse as possible, the FamilieSCN2A Foundation is now providing International Travel Scholarships to both families AND professionals. Apply here!



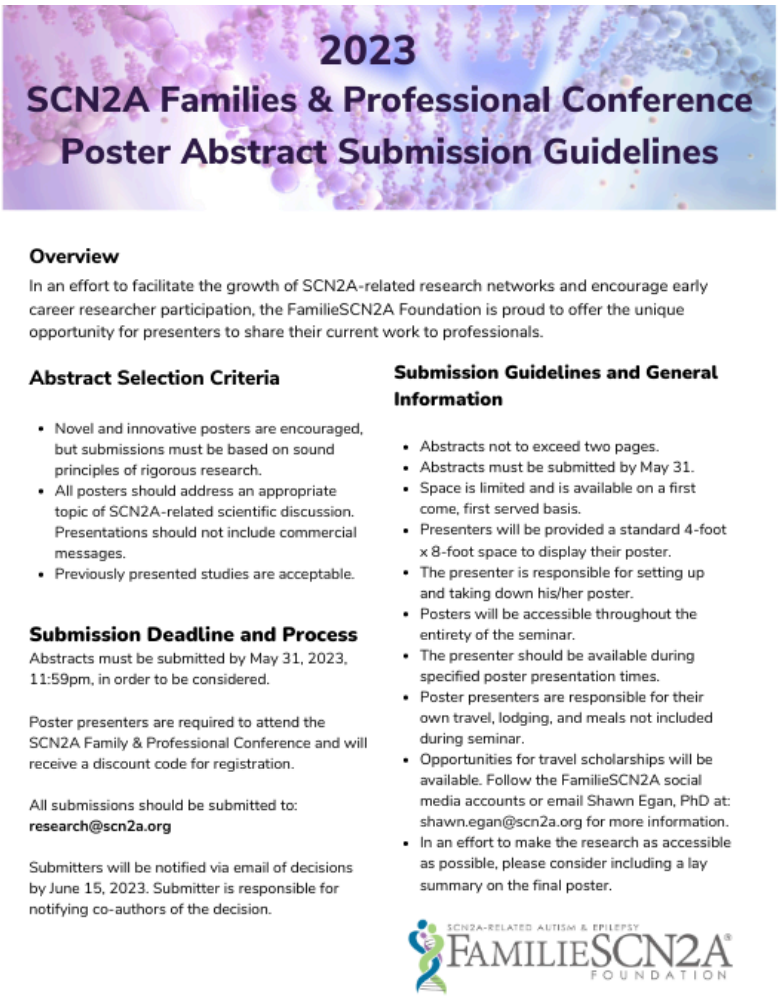
March 8 was a big day for the SCN2A community! TASCO (Team for Accelerating Science and Clinical Outcomes) prepared and Dr. Scott Demerest delivered a moving presentation to the CDC in a public meeting advocating for a specific ICD-10 code for SRD (SCN2A-related disorders). ICD (International Classification of Diseases) codes are very important, especially in rare disease yet most rare diseases are not granted a unique code.

FamilieSCN2A Foundation is excited to offer travel assistance to international families and professionals for the upcoming conference in Denmark. Apply Now! Applications accepted until April 26 or funding runs out.

MONTHLY RECAP

Attention Researchers:
Action Potential Grant LOI
Due APRIL 1
Don't miss out!
Send your LOI today to research@scn2a.org

Click here for more information on becoming a sponsor of the 2023 SCN2A Family and Professional Conference. We expect more than 250 attendees in person and 150 virtually. Summer in Boston will be fun and expensive! We are grateful for all the support we have so far but need more sponsors to offset the costs of this critical gathering of the SCN2A community.



2023
SCN2A Families & Professional Conference
Poster Abstract Submission Guidelines

Overview

In an effort to facilitate the growth of SCN2A-related research networks and encourage early career researcher participation, the FamilieSCN2A Foundation is proud to offer the unique opportunity for presenters to share their current work to professionals.

Abstract Selection Criteria

- Novel and innovative posters are encouraged, but submissions must be based on sound principles of rigorous research.
- All posters should address an appropriate topic of SCN2A-related scientific discussion. Presentations should not include commercial messages.
- Previously presented studies are acceptable.

Submission Deadline and Process

Abstracts must be submitted by May 31, 2023, 11:59pm, in order to be considered.

Poster presenters are required to attend the SCN2A Family & Professional Conference and will receive a discount code for registration.

All submissions should be submitted to: **research@scn2a.org**

Submitters will be notified via email of decisions by June 15, 2023. Submitter is responsible for notifying co-authors of the decision.

Submission Guidelines and General Information

- Abstracts not to exceed two pages.
- Abstracts must be submitted by May 31.
- Space is limited and is available on a first come, first served basis.
- Presenters will be provided a standard 4-foot x 8-foot space to display their poster.
- The presenter is responsible for setting up and taking down his/her poster.
- Posters will be accessible throughout the entirety of the seminar.
- The presenter should be available during specified poster presentation times.
- Poster presenters are responsible for their own travel, lodging, and meals not included during seminar.
- Opportunities for travel scholarships will be available. Follow the FamilieSCN2A social media accounts or email Shawn Egan, PhD at: shawn.egan@scn2a.org for more information.
- In an effort to make the research as accessible as possible, please consider including a lay summary on the final poster.

SCN2A-RELATED AUTISM & EPILEPSY
FAMILIESCN2A FOUNDATION

POSTER SESSION! In an effort to facilitate the growth of SCN2A-related research networks and encourage early career researcher participation, the FamilieSCN2A Foundation is proud to offer the unique opportunity for presenters to share their current work to professionals. Abstracts must be submitted to research@scn2a.org by May 31, 2023, 11:59pm, in order to be considered. Click here for details and scroll down to POSTER SESSION.

Join us on the Million Dollar Bike Ride for Research!

Sign up before **April 1st (TOMORROW) for the EARLY BIRD SPECIAL** with discounted registration and free MDBR bike jersey along with an MDBR t-shirt! Click here for more information and to register.

Not a cyclist or unable to make the trip? Don't worry, you can still help us raise funds for SCN2A research and ride or walk to support our cause! Sign up as virtual cyclist here.

Get your **SCN2A-related Autism Awareness social media frames here!** Just in time for Autism Awareness month. Show the world what's important to you while raising awareness of SRD.

Complete the contact form today

UPDATE YOUR CONTACT INFORMATION! As we chart the course for the future of SRD treatments and cures, the most important waypoint is **YOU**, the patients! **It is *critical* to be able to reach you individually**, by email. Every week, our Chief Scientific Officer fields inquiries on the SRD patient population from biotech and pharmaceutical companies. *We will never give out your personal information without your expressed permission.* **Sign up to be counted!**

Got Swag? SCN2A-related Autism shirts, water bottles, mugs and more are available in the online store.

BEYOND THE DIAGNOSIS

As a talented clinician/researcher who has likely seen more SCN2A patients in the past 10 years than most, **Dr. Heather Olson** was a natural choice to join our Medical and Scientific Advisory Board (MSAB). She splits her time between seeing patients in the clinic (25%) and doing research (75%) and has a real passion for both. **"I love the kids! They steal your heart. They deserve better so I try to provide the best care I can with the current knowledge available while pushing the envelope for better treatments."**

Her caring manner and thoughtful consideration of critical issues are of great value to our mission to accelerate research towards better treatments and cures. One of her current projects is a team effort with colleagues Al George and Ingo Helbig to add an SCN2A 'chapter' to the Cambridge Elements in Genetics in Epilepsy. This all-encompassing review, including videos and advocacy information, will be an amazing resource.

Originally from central PA, medical school at Mayo in MN prepared Dr. Olson for winters in Boston where she's been since her residency at Boston Children's. Now also armed with a degree in epidemiology from Harvard, she is poised to with overlapping do broader work interests, including finding the relative frequency of SCN2A in the population and establishing genotype-phenotype correlations. Some of Dr. Olson's other projects include developing an investigator-initiated clinical

trial of early treatment with the ketogenic diet in genetic developmental and epileptic encephalopathies prior to onset of epileptic spasms; researching visual biomarkers for several genes; and working on gene discovery in the area of inflammatory/febrile seizures. She runs a clinical research team and collaborates closely with basic and translational researchers in her epilepsy genetics research.

One of the many hats worn by Dr. Olson is Director of the CDKL5 multidisciplinary clinic at Boston Children's Hospital. While there are several different models for these so-called 'Centers of Excellence,' they generally involve a patient seeing a variety of specialists in one setting over a short time period, sometimes with an opportunity to participate in research. Coordination is the hard part, and it may be difficult for some families to access this type of care due to travel or financial limitations. For SCN2A-related disorders (SRD) it may make sense to partner with other rare diseases that share similar specialist to create these clinic opportunities.

Dr. Olson first came across SCN2A while doing epilepsy genetics research, particularly on Otahara syndrome, early in her career. She understands very well how far we have come in identifying patients through genetic testing, but how far we still must go in finding appropriate treatments for the many different presentations of SCN2A-related disorders (SRD).

"Receiving a diagnosis of an SRD has a clear impact on a patient and the management of it depends on how a patient is affected by their particular variant." There is not a one-size-fits-all cure, and more research is needed to answer all the questions that arise for precision medicine to be successful. That's one reason it's so important for patients and their families to participate in the process in any way they are able: Fill out a survey, participate in research, *attend the summer conference where you can meet Dr. Olson in person!*

When she's not wearing one of her many professional hats, Dr. Olson may be found chasing her kids around as the family enjoys adventures in the great outdoors – even during cold New England winters.