SCN2A RELATED AUTISM & EPILEPSY
FAMILIESCN2A
FOUNDATION
WWW.SCN2A.ORG

Welcome to our 3rd Quarter Newsletter!
Check out what we have been up to...

SCN2A CONFERENCE UPDATE

We are thrilled to share that the 3rd SCN2A Family & Professional Conference was a smashing success! The weekend was filled with a positive spirit, groundbreaking science, fierce dedication and passion for curing SCN2A disorders. You asked, we listened, and this year we offered two tracks: One geared towards parents and one that was heavier on the science. Each of the conference rooms were well attended with eager learners.

Families who participated are now empowered with education to bring home to their local care teams to ensure their children are on the best possible course

of treatment. Professionals had the rare opportunity to meet children affected by what they are studying in their labs. Through tear filled eyes, they expressed their gratitude to all of the families for the inspiration they gained from listening to their SCN2A Warrior stories. Everyone agrees that a cure is on the horizon and no longer just a dream.

The new Faces of SCN2A video debuted at dinner along with two others. (Many thanks to Kristy Kargel for creating these!) The new videos along with the conference presentations are now available at www.scn2a.org. Or, you may click on the links below:

The Faces of SCN2A - https://vimeo.com/351950742

SCN2A Angels - https://vimeo.com/351950312

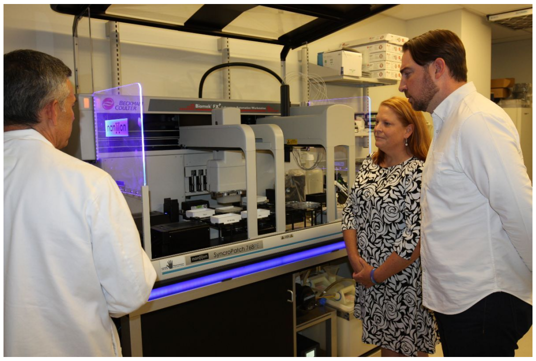
SCN2A Advocacy Video - https://vimeo.com/351948441

Conference Presentations - https://www.scn2a.org/videos.html

For those who attended, PLEASE help us make the next conference even better by sharing your feedback through the Conference Survey link below.

Conference Survey

RESEARCH



Left: Will Hutson, Leah Schust Myers, Dennis Echevarria and Sunita Misra at Northwestern University.

Right: Carlos Vanoye, PhD, shows Leah and Will 'Das Machine.'

Leah Schust Myers, Executive Director, and Will Hutson, Board Member, visited Al George, MD, and his team at Northwestern University last month. Dr. George is the Primary Investigator for the Center Without Walls (CWoW), an interdisciplinary research center dedicated to

advancing the genetic understanding of epilepsy. The CWoW's main focus is investigating sodium and potassium channel genes, the dominant class of genes responsible for early childhood-onset epilepsy. The center is made up of a collaborative group of investigators at five academic medical centers, including Northwestern University, three free-standing research hospitals and two industry partners.

SCN2A is one of the top genes being investigated at the CWoW and Dr. George was proud to give Leah and Will the grand tour of the center. Currently, at least 48 SCN2A variants have been created and studied on a very advanced piece of equipment in the lab called a Syncropatch. This allows for automated electrophysiology of variants at a much faster pace. The machine, nicknamed 'Das Machine,' also looks at pharmacological effects on the variants in high throughput. This is critical to testing existing, as well as new, compounds in variants to determine efficacy.

In addition to creating the variants on Das Machine, SCN2A variants are being studied in iPSCs (induced pluripotent stem cells) and mouse models. This comprehensive approach to looking at the same variants across many models is crucial to understanding the SCN2A gene function and dysfunction and finding new ways to correct it.

The entire SCN2A community is fortunate to have such a dedicated, passionate and brilliant team of researchers focused on helping our kids. Thank you to Al George, MD, Jennifer Kearney, PhD, Anne Berg, PhD, and Sunita Misra, MD, PhD for the amazing work!

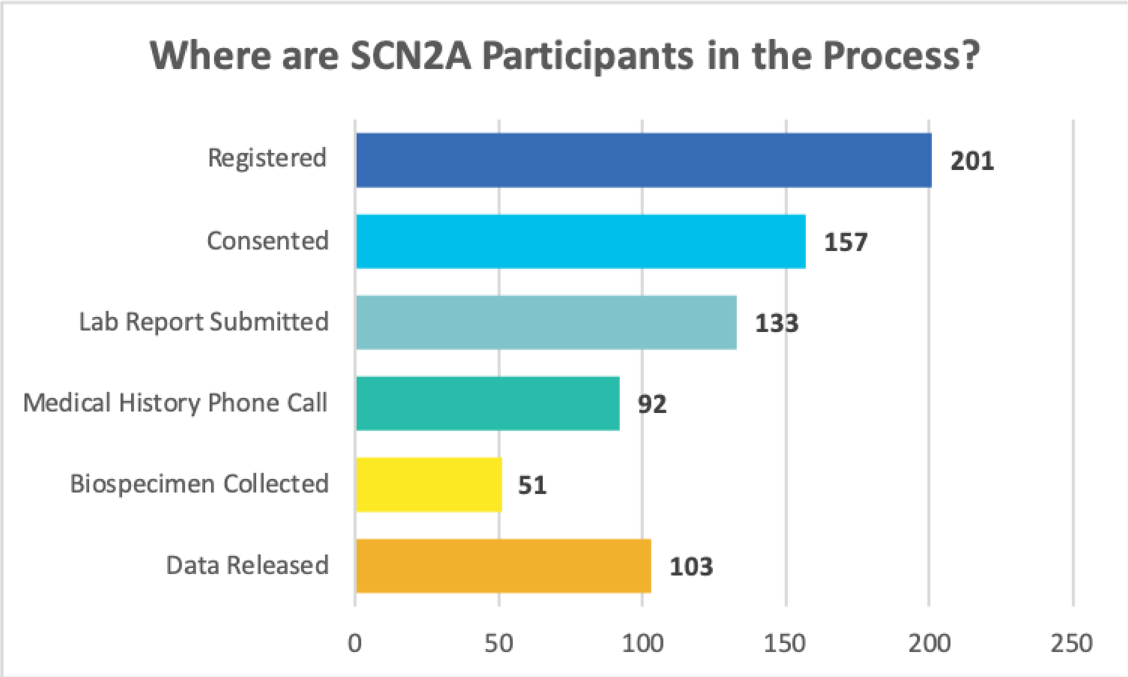
DID YOU KNOW that this year at our Conference we were able to give financial assistance to 18 young investigators, including students and post-docs, to attend? This was made possible through the generous support of the American Epilepsy Society Research & Training Council. The goal of this grant program is to make it easier for young investigators to attend events which provide valuable

opportunities for inspiration that can last an entire career.

"Attending the 2019 FamilieSCN2A Conference in Seattle was a tremendous experience. I had just started working in research, and I was lucky enough to attend [this] as my first conference. FamilieSCN2A tackled a very nuanced gene event with a comprehensive, interdisciplinary approach. As someone who knew very little about sodium channels and their related gene events, the conference provided a top to bottom overview of how the gene works in the body, possible outcomes for those with gene events within SCN2A, and potential treatments in development. Although the research was very stimulating and important, the family testimonials made the biggest impact for me. Having the families share their experiences helped me contextualize the research being presented and helped me realize why I ultimately work in research. Experiencing the worldwide network that FamilieSCN2A has created inspires me to work hard and educate others about SCN2A."

Wesley Ganz (BS Psychology) Research Assistant, Bernier Lab
Career goal: Cognitive Neuroscience Research Scientist

SCN2A continues to lead the way with participation in research with Simons Searchlight. We have 201 individuals with SCN2A registered so far! Click here for the full July 2019 Registry Snapshot.



If you have not done so already, please log in to your account on the new website (simonssearchlight.org) and reset your password. This is VERY important! Unless you activate your account on the new website, you won't be able to finish up any outstanding surveys and yearly follow-up, and they won't be able to let you know about new tasks to complete. If you have any questions about this process, contact the coordinators at coordinator@simonssearchlight.org.

FOR PROFESSIONALS

The FamilieSCN2A Foundation, together with Simons Searchlight, hosts a bi-monthly professionals only collaboration call. These calls have been an invaluable resource to our research and clinician community. Any professional with an interest in SCN2A is welcome to join this call. If you would like to be added to the next call (date TBD) or are interested in presenting your research, please email research@scn2a.org

Did you know we have a page dedicated to Professionals on our web site? Check it out: http://www.scn2a.org/professionals.html

Thank you to the Professional Roundtable participants in Seattle

UPCOMING FUNDRAISING EVENTS

What in the world is an SCN2A 2K?

Between the dates of October 11-31, the FamilieSCN2A Foundation is encouraging you to take *action* in your local area by organizing an SCN2A 2K! Why a 2k vs a traditional 5k/10k? That is easy! 2K = 1.2 miles and the gene SCN2A encodes for the Nav1.2 protein. Besides that cool fact, 1.2 miles is something almost everybody can commit to running, walking, biking or rolling! We call it a Virtual 2K because people can participate from anywhere, including their living room sofa. The idea is to raise awareness and funding in a FUN way.

How do I participate?

Create a team with other local SCN2A families, your friends and family and community members.

FIRST: Create your team here **A team registration link will be created for you and you can share it all over town!**

SECOND: After creating your team, click on Registration**. Each $35 registrant will receive an awesome, first of it's kind SCN2A 2K T-Shirt and a car window sticker to display proudly and spread awareness throughout the world!**

We understand that asking for donations may be uncomfortable. Please always keep in mind that we need a cure for our kids and are doing it for them. Share your SCN2A 2K link widely: to those you know will want to participate in person AND to those who may just want to donate to your team or to the cause. People WANT to help, this is a super easy way to let them!

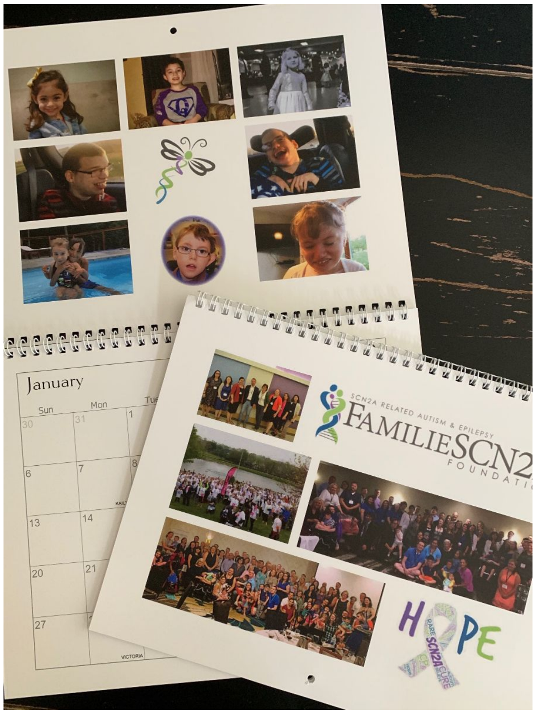
Choose any day from October 11-31 to get together, wear your shirts and any other creative gear and complete the 2K. Please, whatever you do, remember to take LOTS of pictures and post on your social media! This alone will help spread awareness. Use hashtags: #CureSCN2A, #SCN2K, #2K42A

Contact us at info@scn2a.org if you would like help setting up your 2K.

Get started TODAY!

FOR FAMILIES

To celebrate National Grandparents Day we announced the FamilieSCN2A Grandparents Club! This is an effort brought forward by the amazing grandparents at the recent SCN2A Family and Professional Conference who saw a need for supporting one another on the journey of grandparenting kids with special needs. To learn more and get involved please email Ellen Kaplan: ellen@possibilities-at-work.com and/or Suzanne Schust: suzschust@gmail.com.



CALLING ALL SCN2A MODELS!

Please help us raise awareness and funds by adding a photo of your beautiful SCN2A warrior to the 2020 FamilieSCN2A calendar. First time participants: Email a high quality photo, your child's birth day and month (not year) and that you give permission to publish it to Carla.Forbes@scn2a.org. If you have already sent a photo for the birthday club, please indicate that in your email. Previous calendar models: Let Carla know if you would like your child to be on the 2020 calendar. If you would like to send a new photo, please do!

The deadline is OCTOBER 1ST

Calendars will be ready to order in time for holiday gift giving.

HOW CAN I HELP?
There are many ways to contribute to the FamilieSCN2A Foundation! To volunteer your time and talents please email info@scn2a.org. To learn more about Planned Giving, Stock Donations, Used Car Donations, Amazon Smiles and other ways to donate, visit www.scn2a.org/donate
We can now receive donations via text! Text "CURESCN2A" to 443-21

We are pleased to announce that Michael Vasey has joined the FamilieSCN2A Foundation Board of Directors as Treasurer.

Michael believes every child should have an opportunity to achieve a happy, fulfilling and productive life. Although Michael does not have a special needs child, he believes helping FamiliesSCN2A Foundation achieve its mission will make the world a better place. Assisting the Foundation to build awareness, provide support, and find a cure for children with SCN2A is the ultimate reward. Currently, Michael is the Chief Financial Officer of Fortiss, LLC and serves as a Director on five California cardroom entities in the State of California. Michael earned an undergraduate degree in Business/Economics from University of California, Santa Barbara, and a Masters of Business Administration from the Loyola Marymount University. Additionally, Michael is a licensed Certified Public Accountant in the State of California.

We are pleased to announce that Amy Kozsuch has joined the FamilieSCN2A Foundation Board of Directors as a Director-At-Large

Amy has over 20 years of experience leading clinical teams, healthcare executives and physician practices to drive positive change in care delivery systems and overall healthcare markets. She has developed and enhanced integrated networks of care, strategic alliances, physician and executive leadership relations, rural health and health education. In addition to service to our board, Amy is the president of the National Kidney Foundation of Florida, past president of the Florida Renal Association and a former DaVita executive in the areas of Business Development and Kidney Care. Amy earned her MSN, MBA and HCA from University of Phoenix, where she is currently a doctoral candidate.

"Nothing great was ever achieved without enthusiasm" - Ralph Waldo Emerson

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HELP US SPREAD AWARENESS!
Have you seen our SCN2A apparel? You can now order from our apparel shop year round! The apparel comes is sizes ranging from youth to 3XL depending on the product you order. Apparel ships internationally too! Click on the photo to visit the shop. Thank you for supporting the FamilieSCN2A Foundation and helping us get one step closer to effective treatments and a CURE! https://www.bonfire.com/store/scn2a-awareness-apparel/

CONNECT I SUPPORT I RESEARCH

Putting SCN2A on the map!

Our mailing address is:
P.O. Box 82, East Longmeadow, MA 01028