



Welcome to our 2nd Quarter Newsletter!  
Check out what we have been up to...

SCN2A CONFERENCE UPDATE



JOIN US IN SEATTLE!

AUGUST 1 - 3, 2019

## THE SCN2A FAMILY & PROFESSIONAL CONFERENCE

**LOCATION:** Graduate Hotel Seattle  
4507 Brooklyn Avenue N.E., Seattle, Washington, 98105

**REGISTRATION:**  
Thursday August 1st from 4:00 p.m. - 7:00 p.m.

**CONFERENCE RUNS:**  
FRIDAY August 2nd 8:00 a.m. - 5:00 p.m.  
SATURDAY August 3rd 8:00 a.m. - 2:30 p.m.  
(families) and 3:00 p.m. (professionals)

The cost of registration for the 2019 conference is \$175 for families which includes up to 6 participants total. The cost for professionals in the industry is \$250 or \$300 for those participating in the CME course. All pricing includes entry into all sessions Friday - Saturday, breakfast and lunch both days, the SCN2A Family & Professional Appreciation Dinner on Friday August 2nd and any organized activities.

BROUGHT TO YOU BY:



For more information or to REGISTER visit:  
[www.scn2a.org/conference.html](http://www.scn2a.org/conference.html)



The University of Washington School of Medicine is accredited by the Accreditation Council for Continuing Medical Education to provide continuing medical education for physicians.

The University of Washington School of Medicine designates this live activity for a maximum of 12.5 AMA PRA Category 1 Credits™. Physicians should claim only the credit commensurate with the extent of their participation in the activity.

### NOW OFFERING CONTINUING MEDICAL EDUCATION (CME) CREDITS!

Are you a medical professional considering attending the SCN2A Family & Professional Conference in Seattle, Washington? Now you can earn CME credits by attending! If you have already registered and would like to upgrade your registration to include the Continuing Medical Education (CME) course, please email [info@scn2a.org](mailto:info@scn2a.org). If you have not registered yet there are still some slots available. Click on the purple Register Now button below.

We are still accepting [Exhibitors](#) and Sponsors for the conference, including the opportunity to sponsor a family's attendance. Details available at the lower section of our

**Families it's not too late to register, there still is space!**

Hotel rooms are limited so we recommend booking a room at the [Graduate Seattle Hotel](#) as soon as possible. To book your room email Reva Cloy [rcloy@graduateseattle.com](mailto:rcloy@graduateseattle.com) and mention "Group Code": 190801FAMI

**Register Now**



Share



Tweet



Forward

---

### **Here's a sneak peak at some of the agenda topics for the SCN2A Family & Professional Conference:**

- How Sodium Channels Work
- SCN2A 101: Functions, Mutations, and Disease
- How to read your genetic report and what you NEED to know
- What happens when there is a change in the SCN2A Gene
- Clinical Presentation of SCN2A and The difference between Epilepsy Syndrome and Genetic Etiology
- Hot, Stiff, Twitchy, and Shaky: It is not a seizure, but it is NOT normal
- Clinical Phenotype of Autism and ID in SCN2A
- SUDEP and Mortality in SCN2A
- Exploring unusual aspects of SCN2A mutations: gating pore currents and response of SCN2A resurgent currents to CBD
- Expanding the Functional Repertoire of SCN2A Variants

## **RESEARCH**

Thank you to our donors, SCN2A families and friends who have raised money for the FamilieSCN2A Foundation! Many of those donations have been allocated to fund our investigator-initiated

grant program: **take ACTION, create POTENTIAL.**

[Click to read the Press Release](#)

**take  
ACTION,  
create  
POTENTIAL**

**2019 Grant**



**Recipient**



The FamilieSCN2A Foundation is pleased to announce Misra Sunita as one of the recipients of the 2019 Action Potential Grant. Misra has provided a summary of her proposed work:

SCN2A dysfunction changes the way brain cells communicate both electrically and chemically leading to epilepsy and neuropsychological comorbidities. I will use EEG to identify seizure patterns and the role of sleep on seizures in a new mouse model of SCN2A-related epilepsy. Then I will look at neurotransmitter levels in the brains of mice with SCN2A-related epilepsy. I will use drugs that target the abnormal neurotransmitter levels to improve abnormal electrical and chemical signaling in the brain. This work may identify new druggable targets for better control of epilepsy and associated comorbidities in SCN2A-related epilepsy.

I began my professional research career studying SCN2A-related epilepsy in 2002. With this project, I feel like I have come full circle to pursue my passion. Together we can deepen our understanding and look for new therapeutic targets to help children with SCN2A-related disorders. Thank you FamilieSCN2A Foundation for awarding me the Action Potential Grant!

# take ACTION, create POTENTIAL

2019 Grant



Recipient



The FamilieSCN2A Foundation is also pleased to announce Caitlin M. Hudac, Ph.D., Research Scientist, The Bernier Lab, University of Washington, as a grant recipient.

Caitlin's research is on SCN2A Neural Biomarkers of Attention. Aligned with the #FamilieSCN2AFoundation mission to improve the lives of those affected by SCN2A-related disorders, this project aims to generate a candidate biological indicator ("biomarker") that can be used to track changes in children with SCN2A disruptive mutations. This will be critical for developing and assessing the effectiveness of clinical interventions. For this project, 20 children with disruptive SCN2A mutations will wear an electroencephalography (EEG) net while watching movies. We will test an auditory attention brain biomarker and characterize how these brain responses to sounds relate to other aspects of the child's behavior.

.....

# SIMONS SEARCHLIGHT

Driven by science. United by hope.

Did you know Simons VIP has changed its name to Simons Searchlight? They did this to reflect their mission of working together with foundations like ours to find answers. Along with the new name, they updated their website with a more user-friendly registration process, making it easier to join and access useful features. Their focus continues to be catalyzing research to drive the science of autism-related genetic changes forward.

Simons Searchlight is still the official, central SCN2A registry. As a foundation we partnered with Simons years ago to collect longitudinal data that defines the disorder. Simons makes the de-identifiable data available to qualified researchers around the world to expedite finding treatments and a CURE and will continue to do so. The more researchers looking at this disorder from multiple angles the better. Is your family included? Start the process today! Simons Searchlight rewards you for your time and effort with gift cards!

<http://www.simonssearchlight.org>

#bepartofthecure #rare #SCN2A #CureSCN2A

---



Having a child with an SCN2A disorder is challenging on more levels than one can ever explain. Most of the warrior caregivers would agree that imagining the “what if” is the most difficult. FamilieSCN2A Foundation and Simons (SFARI) endorse the Autism BrainNet as the official repository for the ultimate gift of donation. You can read more about the impact the Autism BrainNet is making to the field

here: <https://autismsciencefoundation.org/news/autism-brainnet-makes-resources-available-scientific-community/>

"The human brain is the most complex and unique object in the known universe. Our attempts to understand how it works, and how it goes wrong in human disease, are hindered by our inability to test human brain tissue directly. Instead we rely on less complex animal brains or small numbers of human brain cells grown in a dish. Some of the data we generate from these is a good reflection of how the human brain works, but other data is not. Even a small number of human brain samples provides critical information to allow us to distinguish the good and informative data from the poor and irrelevant data. Having access to these human samples is like a navigator glimpsing the north star, providing a point of truth in the dark to guide them home. The brave and generous donations these families make will have far reaching and important impacts in our quest to develop future therapeutics. " - Stephan Sanders, BMBS, PhD <https://humangenetics.ucsf.edu/sanders-stephan/>

***“Amelia always fought for a cure for SCN2A. She continues to do so now, even after her death, with the gift of her donation. Amelia’s gift is important because it furthers SCN2A research towards our ultimate goal- a cure for SCN2A.” - Angie Weaver, Mom to Amelia***

---

## 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. Our next call is scheduled for **June 25 from 12:00 to 1:00 Eastern**. Any professional with an interest in SCN2A is welcome to join this call. If you would like to be added to the call, please email [research@scn2a.org](mailto: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>

---

## UPCOMING FUNDRAISING EVENTS



On June 15, Eliana Lewis and her Force will be hosting the 4th Annual Eliana's Force Golf Tournament supporting The FamilieSCN2A Foundation.

If you are in Central Missouri, sign up to play or just swing by to say hello. If you would like to join Eliana's Force, but cannot make it to the tournament, you can



do it virtually by going to Eliana's FirstGiving Page:

<https://www.firstgiving.com/fundraiser/michelle-lewis-6/elianasforce>

---



For the second year in a row, Jamie Tuminello (mom of Marty) is graciously supporting the FamilieSCN2A Foundation.

On October 19, 2019, team 'Marty's Marvels' will be running in the Baltimore Running Festival to support kids, like Marty, and their families affected by a change in the SCN2A gene.

Jamie commented: "The FamilieSCN2A Foundation has been our number one resource. We hope that you join us and support kids like Marty by donating to our run for the FamilieSCN2A Foundation."

Join Marty's Marvels and support the FamilieSCN2A Foundation by going to:

<https://www.firstgiving.com/fundraiser/jamie-tuminello/MartysMarvels>

---

FOR FAMILIES



**A New Faces of SCN2A Video Project Is Underway!**  
**We are still in need of photos!**

We are in the process of creating an updated **Faces of SCN2A video** which will premier at the SCN2A Family & Professional Conference August 1st – 3rd. This is the **last call** for photos and we need your help! If you would like to participate in this project please send the following to [marketing@scn2a.org](mailto:marketing@scn2a.org) no later than **June 30th**:

1. Full size photo of your child(ren) who have been diagnosed with SCN2A
2. A photo of your *favorite* piece of artwork your child has created. It can be any media, just snap a photo of it and send it to us! (optional)
3. A current family photo
4. Your child's variant (optional)

Please note in your email:

*"I, (state your name) give permission to the FamilieSCN2A Foundation to use these photos on their website, social media outlets and in any marketing materials."*

To view the 2017 Faces of SCN2A video scroll down to the grey section of our video page: <https://www.scn2a.org/videos.html>

Questions? Contact Kristy Kargel at [marketing@scn2a.org](mailto:marketing@scn2a.org)

*Artwork shown was created by Erik P.*

---

## HELP US SPREAD AWARENESS!

Have you seen our SCN2A apparel?  
You can now order from our apparel  
shop year round! The apparel comes  
in 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/>



## Let's Celebrate!



*Enjoy your special day!*



## JOIN OUR BIRTHDAY CLUB!

As a kid, it's so much fun getting mail  
addressed just to you! Sign up today  
for our Birthday Club and your child  
will receive a birthday card from  
us. With your permission, we will  
even do a special shout out posting  
on social media on the actual  
birthday (this part is optional and not  
required to join the Birthday  
Club).

[Click Here to sign up!](#)

---

