



Welcome to our 4th Quarter Newsletter!  
Check out what we have been up to...

## RESEARCH ADVANCES

### **Announcing the First Phase 2 Clinical Trial for the Treatment of Rare Epilepsy Including Sodium Channels Such as SCN2A**

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*Over the last four years, FamilieSCN2A Foundation Board members have served in the Patient Advisory Group for Zogenix Inc., a global pharmaceutical company. They are a patient focused team who care deeply for the communities they serve.*

[Zogenix](#)'s application for their investigational drug FINTEPLA in Dravet syndrome is now under review by the FDA, with a target approval date of March 25, 2020. Top line data for FINTEPLA in LGS is expected during Q1 2020. On their recent 3rd quarter conference call, Zogenix announced a new Phase 2 study of FINTEPLA in multiple rare epilepsy disorders to understand various characteristics of these disorders and evaluate whether fenfluramine is safe and effective in these patient populations. Indications will include Doose syndrome, tuberous sclerosis complex, Dup15q syndrome, CDKL5 deficiency disorder, mutations in PCDH19 gene, mutations in Na<sup>+</sup> channel genes that do not meet diagnostic criteria for Dravet syndrome, and Dravet syndrome patients aged 1 to <2 years old only. Expected enrollment to begin in the first quarter of 2020. Inclusion/exclusion criteria as well as participating clinical sites to be announced soon.

For more information email [research@scn2a.org](mailto:research@scn2a.org)

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FamilieSCN2A is proud to announce the support of a collaborative research project in Dr. Kathrin Meyer and Dr. Nicolas Wein laboratories at Nationwide Children's Hospital, Columbus, Ohio, USA. Dr. Meyer's laboratory concentrates on disease modeling of neurodegenerative and neurological diseases with a strong translational focus. Moreover, Dr. Meyer and Dr. Wein develop gene therapies for neurological and neuromuscular disorders using small RNA-based approaches to increase full length protein expression or decrease production of toxic RNAs. Dr. Meyer and Dr. Wein are characterizing multiple SCN2A mutation carrying cell lines that have been collected from patients over the last year. They will develop a robust assay for drug screening and testing of new therapeutic strategies using these cells. They will also perform expanded compound testing and work on understanding underlying disease mechanisms in more detail. Finally, they will develop and test gene therapy approaches on the different patient cell lines.

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**Foundation leaders participated in the Center Without Walls (CWoW) annual 'in person' meeting at AES. With a packed agenda, the team reported updates and progress made in their first year. It was an honor to be included in this meeting to represent the patient community. An exciting update to share is the creation**

of the website for the [Channelopathy-Associated Epilepsy Research Center \(CAERC\)](#). Check out the “Tools” section to see details on SCN2A variants. As the CWoW studies and classifies each variant, the information will be made public on their site.

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[Xenon Pharmaceuticals](#) recently announced a license and collaboration agreement with [Neurocrine Biosciences](#) to develop first-in-class treatments for epilepsy. Neurocrine Biosciences gains an exclusive license to XEN901, a clinical stage selective Nav1.6 sodium channel inhibitor with potential in SCN8A developmental and epileptic encephalopathy (SCN8A-DEE) and other forms of epilepsy, including focal epilepsy. In addition, Neurocrine Biosciences gains an exclusive license to pre-clinical compounds for development, including selective Nav1.6 inhibitors and dual Nav1.2/1.6 inhibitors. The agreement also includes a multi-year research collaboration to discover, identify and develop additional novel Nav1.6 and Nav1.2/1.6 inhibitors. <https://www.prnewswire.com/news-releases/neurocrine-biosciences-and-xenon-pharmaceuticals-announce-agreement-to-develop-first-in-class-treatments-for-epilepsy-300967168.html>

***Because of this partnership, Xenon will now have the resources to move this small molecule treatment to clinical trials much faster. This research is important for our community because SCN2A encodes the Nav1.2 channel and there are similarities in presentation of SCN2A and SCN8A such as early seizure onset & DEE.***

Feedback from the FDA suggests that Xenon may be able to advance XEN901 directly into a Phase 2, or later stage, clinical trial examining efficacy in pediatric patients with SCN8A-EE, without requiring an adult clinical trial first. Xenon is currently in the process of completing a pediatric formulation of XEN901 and juvenile toxicology studies to support pediatric development activities. Xenon intends to run a Phase 1 pharmacokinetic (PK) study in adults with the new pediatric formulation soon and then initiate a Phase 2/3 clinical trial in SCN8A-EE patients. Details about the final trial design, anticipated timing, criteria, and endpoints will be disclosed over the coming months.

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**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. Please read below for progress reports. In 2020, our goal is to fund three more **Action Potential** Grants.

Your continued support is vital to our mission to find better treatments and a CURE for SCN2A Disorders.

Donate

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**take  
ACTION,  
create  
POTENTIAL**

**2019 Grant**



**Recipient**



In the past, anticonvulsant medications have not been tested for seizure reduction in mouse models of genetic epilepsy. Instead pre-clinical medication trials look at reduction of seizures induced with chemical or electrical stimulation. Recently [Dr. Jennifer Kearney's lab](#) at Northwestern University made a new mouse mode of SCN2A-related epilepsy. As part of her FamilieSCN2A Foundation Action Potential Grant, Dr. Sunita Misra is studying seizures in this new mouse model. Sunita's ongoing work shows that seizures in the new Scn2a mouse model are different than seizures caused by chemical or electrical stimulation. These differences may partly explain why many children with SCN2A-related early onset epilepsy have seizures that are difficult to control with current anticonvulsant medications. Further work will look at the effectiveness of FDA approved anticonvulsant medications at blocking seizures in the Scn2a mouse model.

# take ACTION, create POTENTIAL

2019 Grant



Recipient



Research is a GO for Dr. Caitlin Hudac at the University of Alabama! Over the past 4 months, Dr. Hudac has obtained research approval, set up new equipment, and established her [Brain Research Across Development \(B-RAD\) Lab](#)! She has 14 undergraduate research assistants working hard to learn about the brain and SCN2A. They are now scheduling participants to study and have slots available for any eligible SCN2A families who would like to participate—click on the lab website link above for more details!

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AMERICAN EPILEPSY SOCIETY MEETING





Michelle Lewis, Jenny Burke, Amy Richards, Leah Schust Myers, Carla Forbes  
advocate for SCN2A at the 2019 AES Annual Meeting in Baltimore, Dec 6-10.

At the world's largest gathering on epilepsy, representatives of the Foundation spent several days having hundreds of conversations and distributing materials to educate and spread awareness to clinicians, many with SCN2A patients, as well as research scientists, industry leaders and other patient advocacy organizations. Time out of the exhibit hall was spent in meetings and workshops to push our mission forward, encouraging collaboration and cooperation as we strive for better treatments and a cure. Highlights include:

- Meeting with multiple Industry leaders to discuss current and future partnerships.
- Hosting a collaborative meeting including all stakeholders to define a clear strategy to harmonize all existing SCN2A data in one central location accessible to all. Most importantly, data will be accessible to patients, reducing the need to reenter the same information into multiple systems.
- Participating in a planning committee meeting for the next Sodium Channel Coalition (SCC) conference.
- Partnering with the American Epilepsy Society (AES) to fund a young investigator grant specific to SCN2A research.

- Attending a training workshop with the Child Neurology Foundation on peer-to-peer support. A great deal was learned about ways to better serve our community and we are excited to launch some new programs in 2020.
- Collaborating with leaders of other rare epilepsy groups where we redefined the role and identity of the Rare Epilepsy Network (REN).
- Discussing many collaborative efforts in the landscape of epilepsy and rare disease as members of the Epilepsy Leadership Council.



One of our favorite things at AES was organizing our annual genetic epilepsy dinner with over 60 attendees including patient advocates, clinicians, industry reps and scientists. This was a fun and casual get together for socializing and getting to know one another on a personal level.

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## NEW PARTNERSHIPS



FamilieSCN2A Foundation is proud to be one of the founding members of DEE-P (Developmental Epileptic Encephalopathy-Project) Connections. DEE-P Connections came about as a way to break through the isolation of having a child with profound, complex needs, to facilitate connections and share critical resources with families facing similar challenges. Check out the upcoming webinars: <https://www.deepconnections.net/upcoming-webinars>

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The FamilieSCN2A Foundation has joined forces with 12 other neurodevelopmental disease organizations to launch [COMBINEDBrain](#). This is a collaborative effort to develop outcome measures and biomarkers and fast track the drug development process. Terry Jo Bichell, PhD is a fearless leader in this space. We have a busy year planned and the timing for the SCN2A community could not be any better as multiple new treatments are in the pipeline.

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**NEW: THE SCN2A ONLINE STORE!**



## HELP US SPREAD AWARENESS!

The 2020 SCN2A Little Warriors Calendar is here! It's available in two sizes and can be shipped internationally. And, you can now order from our updated apparel shop year round! Click on the photos to reach the shop which can also be accessed using the [STORE button](#) in the top right corner of [www.scn2a.org](http://www.scn2a.org).



**International SCN2A Awareness Day is February 24.** One easy way to raise awareness is to post pictures on social media of your family and friends in SCN2A gear! Place your order and then plan a gathering with other SCN2A families in your area on or near February 24. Did you know you can apply for a [Family Event Grant](#) to help with the cost? Questions? Email [community.support@scn2a.org](mailto:community.support@scn2a.org).



*Whether we're working at home alone while juggling our kids or caught up in the whirlwind of the AES meeting, we never forget who we're fighting for and how far we have come since the first AES meeting we attended 4 years ago.*

*-Your FamilieSCN2A Board of Directors*

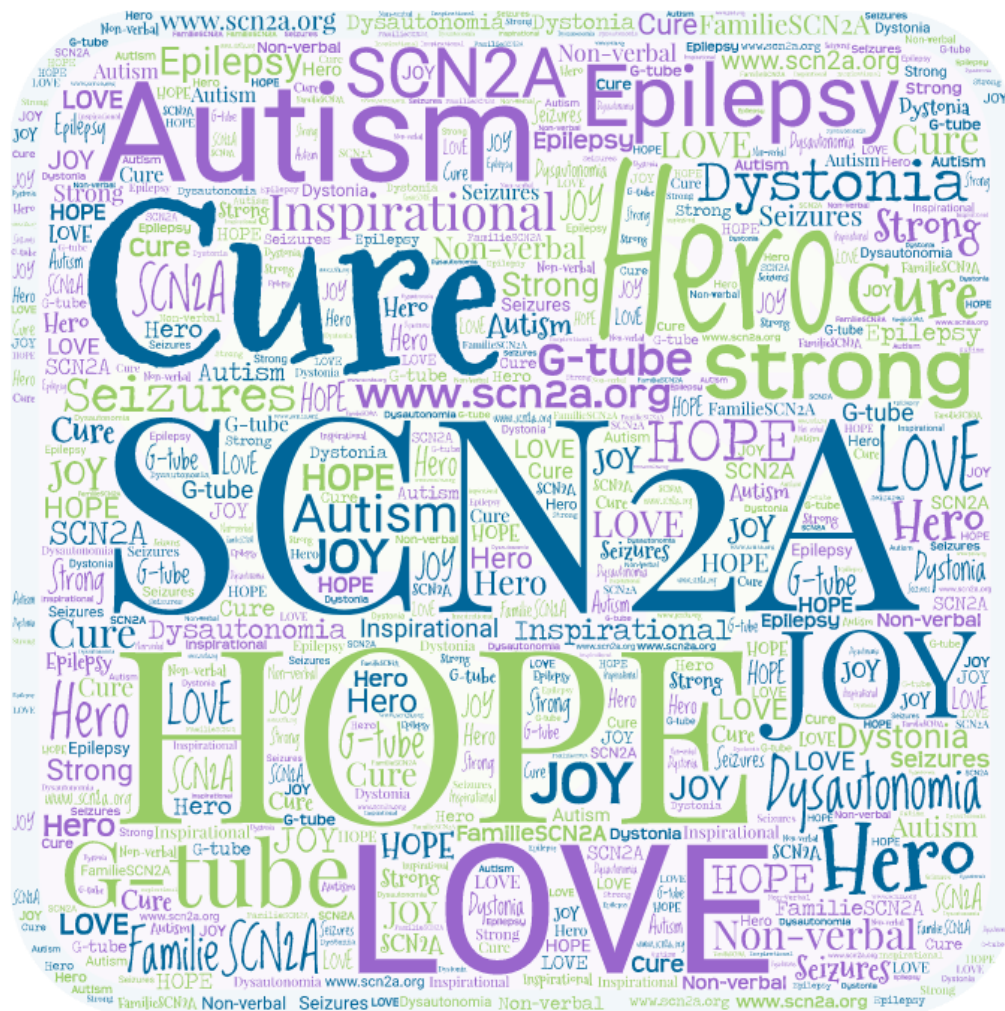
Our **vision** is to find effective treatments and a cure for SCN2A related disorders. Our **mission** is to improve the lives of those affected by SCN2A related disorders through research, public awareness, family support and patient advocacy.

To accomplish our vision and mission, we will:

- Coordinate and collaborate with the global scientific community to understand the function of the SCN2A gene in order to develop effective

treatments and a cure for SCN2A disorders

- Increase medical community and public awareness of the complexity and potential severity of SCN2A disorders
- Provide educational and emotional support for those affected by SCN2A disorders
- Raise money to fund our goals



Our mailing address is:

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