



## Building Community

We are thrilled with the astounding response to attend the SCN2A Summer Seminar in person this year! The hotel is SOLD OUT and we have very limited space still available nearby.

**In-person registration closes on June 17th at 5 pm ET.**

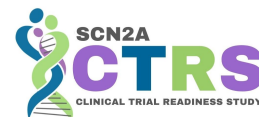
Do not delay, [REGISTER TODAY](#) to attend in person or  
[Register here to join via Zoom](#)

#SCN2A #SCN2ASummerSession2022



## Building Research

**Participants are starting to cross the finish line and researchers are already asking for the data!** Thank you for your participation in this important study. **Please** take the time to keep up with your surveys and interviews by logging into [SCN2ACTRS.study](https://scn2actrs.study). For questions, contact [SCN2ACTRS@SCN2A.org](mailto:SCN2ACTRS@SCN2A.org).  
**The SCN2A community is READY for clinical trials!**



## Early CTRS Results

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**Coming soon to our website:** Updates designed to educate the SCN2A community about research and to provide resources for making informed decisions about participating in clinical trials. For instance, there is a lot of excitement and confusion around clinical trials, but what does that really

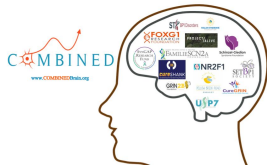
mean? Medical research studies involving people are called clinical trials. There are two main types: Interventional and observational.

**Interventional trials** aim to find out more about a particular intervention, or treatment.

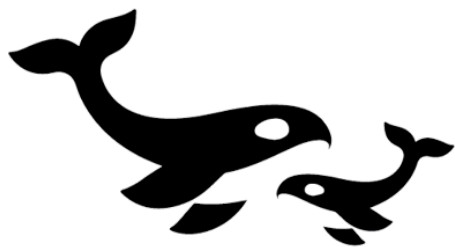
**Observational studies** aim to find out what happens to people in different situations.

For our purposes, we will use [NIH definitions](#) and call an interventional study a Clinical Trial. If a project is observational or information-gathering, we will call it an Observational or Research Study.

In the meantime, check out this excellent [toolkit](#) created by our rare disease partners at Global Genes. We are grateful for their support!



The FamilieSCN2A Foundation is excited to be included in a grant that Duke University and COMBINEDBrain have received from the FDA for a tool to measure communication ability, called the **Observer-Reported Communication Ability (ORCA)** measure. [Press release.](#)



**ORCA**

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Observer-Reported Communication Ability Measure

## **New interview study seeking caregivers of children with SCN2A related disorders**

We are looking for caregivers of children with SCN2A to participate in a 1 hour phone interview to discuss their child's communication abilities and complete and provide feedback on a communication survey we've designed.

**You may be eligible for this research study if you are:**

- A caregiver or parent of a child between the ages of 1 and 18 with a diagnosis of SCN2A
- Are able to speak and read English
- Are able to participate in a 1 hour interview conducted via phone, Zoom or Webex

Interviews will be recorded and transcribed. Participants will be compensated for their time.

**For more information please contact:** [EORCAStudy@dm.duke.edu](mailto:EORCAStudy@dm.duke.edu)

**We are excited to announce two new opportunities for the SCN2A community!**

*Any specific company, products, processes, or services by trade name, trademark, manufacturer, or otherwise on the website or social media platforms does not constitute or imply the FamilieSCN2A endorsement, recommendation, or favoring by the Foundation. We encourage any interested participants to research, consult with your doctor, ask questions, and get input from multiple, unbiased resources.*



**Now enrolling: the PACIFIC study for the treatment of adults with developmental and epileptic encephalopathies**

Frequent epileptic activity associated with developmental and epileptic encephalopathy (DEE) can have a profound negative impact on brain development. This may cause motor impairment and may have a large impact on patients' quality of life.

Longboard Pharmaceuticals, Inc. is studying an investigational drug for patients who have DEE. The purpose of this clinical study is to test the safety and tolerability of multiple doses of the investigational drug, LP352 and find out how LP352 works when given in addition to antiseizure medication in adults with DEE. This study will consist of 10 to 11 visits over a period of 22 weeks.

**Participants may qualify for this study if they:**

- Are 18 to 65 years of age
- Have been diagnosed with developmental and epileptic encephalopathy (DEE)
- Are currently taking 1 to 4 antiseizure medications at a stable dose
- Have a reliable caregiver

**To learn more about the PACIFIC study, visit:** [www.pacificclinicalstudy.com](http://www.pacificclinicalstudy.com)

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**We're excited to announce** a new development related to our planned EMBRAVE study (PRAX-222), known as EMBRAVE, which is specifically designed for early onset children with gain-of-function SCN2A epilepsy. EMBRAVE (PRAX-222) is planned to launch later this year, and eligibility for that study may require three months of recent electroencephalogram (EEG) data as well as seizure diaries. Patients residing the United States with early-onset SCN2A-DEE now have an opportunity to participate in a preliminary assessment of their condition called the SCN2A EEG Observational Study, which involves at-home collection of this information that could provide information that may facilitate later enrollment in EMBRAVE (PRAX-222).

**Who is eligible to participate in the study?**

Males or females, ages  $\geq 1$  to  $\leq 16$  years old

Residents of the United States  
Diagnosed with SCN2A variant through genetic testing  
Had seizure onset within the first month of life  
Had 8 or more motor seizures in the last 4 weeks

### What does participation involve?

During the study, information such as EEG data and seizure diaries will be collected fully remotely (i.e., in the home setting) at specific time points. Enrollment in the SCN2A EEG Observational Study is not a prerequisite for entry into EMBRAVE (PRAX-222). However, data from the Observational Study may facilitate later enrollment into EMBRAVE.

**To learn more and see if your child may qualify, visit:**

<https://scn2a.com/clinical-research-studies/scn2a-eeeg-observational-study/>

*We are Praxis Precision Medicines. We are a clinical-stage biopharmaceutical company translating genetic insights into the development of therapies for patients affected by central nervous system (CNS) disorders characterized by neuronal excitation-inhibition imbalance, including patients and families impacted by rare genetic epilepsies. Our dedicated Epilepsy Franchise is comprised of neurologists, geneticists, scientists, and parents of children with SCN2A mutations. And like all parents, we are fighting to give our children the most fulfilling existence we can.*

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## Families: Got CRID?

The FamilieSCN2A Foundation encourages all patients enrolled in research to create a CRID (Clinical Research ID.) This is a **free** service that allows the patient to take control of their own data. By sharing your CRID, researchers can then reuse, merge and share your research data without using your PII (Personal Identifiable Information) or PHI (Personal Health Information).

**For more info and to create your CRID today:** <https://thecrid.org/>



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## SCN2A in the news

Recent publications from our research community:

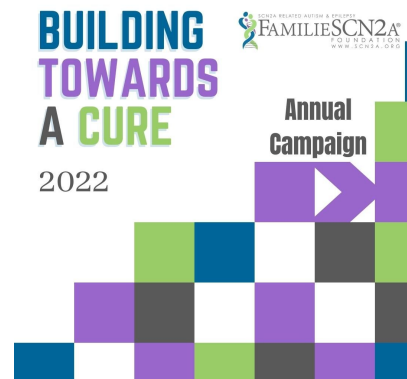
[Caregiver assessment of quality of life in individuals with genetic developmental and epileptic encephalopathies](#), Dev Med Child Neurology, Jan. 26, 2022

[CRISPR activation rescues abnormalities in SCN2A haploinsufficiency-associated autism spectrum disorder](#), BioRxiv, April 1, 2022

[Deficiency of autism-related Scn2a gene in mice disrupts sleep patterns and circadian rhythms](#), Neurobiology of Disease, June 15, 2022

## Building Resources

We fell short of our Annual Campaign goal, but are very grateful to those who donated to raise \$53,862! Thanks to you, we are steadily building towards a cure. While we work to increase our infrastructure with the *CZI Rare As One* grant, we still need to fund research. Your donations will be put to work right away as the next round of [Action Potential Research Grants](#) will be awarded on July 1. [It's not too late to Give Today!](#)







**PLEASE JOIN US on the  
Million Dollar Bike Ride for rare disease research!**

*We need a few more riders and volunteers* to come to Philadelphia and ride with FamilieSCN2A founders, Leah Schust Myers and Carla Forbes, on Saturday, June 11th—or ride virtually from home! [LEARN MORE](#)

**We need a cure NOW so our goal is to maximize the \$30,000 that will be matched by the Orphan Disease Center for SCN2A research!**

Donate to SCN2A Warrior Riders



**SAVE THE DATE!**

THE 2ND ANNUAL  
'GOLF FORE SCN2A'

**AUGUST 29th**  
in BUFFALO, NY!

Email [info@scn2a.org](mailto:info@scn2a.org) for details





**THANK YOU** to all who volunteer to participate in research studies. We know it's not always easy but research is the way to change the future for those currently suffering from SCN2A-related disorders and the ones who will come after us. You give us HOPE.

**Build Resources, Donate Here**

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