We are an organization created by parents of children diagnosed with a rare form of epilepsy and autism as a result of a change in the SCN2A gene.

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.
Our "strategic quadrad" represents four key focus areas aimed at improving the lives of those affected by SCN2A related disorders.
The FamilieSCN2A Foundation funds research to better understand SCN2A, find effective treatments and work toward finding a cure. We are interested in supporting research that advances understanding of the cellular, molecular, genetic and systems-level mechanisms of SCN2A-related disorders. We do this through:

**The FamilieSCN2A Action Potential Grant**

The Action Potential Grant is an investigator-initiated grant program, intended to accelerate the development of therapeutic treatments and disease-modifying advancements for those living with changes in the SCN2A gene. This grant program is designed to facilitate preliminary investigations that will potentially lay the groundwork for subsequent grants from the government, industry, or other funding sources, including the FamilieSCN2A Foundation.

**AES Junior Investigator Research Award**

A full research grant will be funded by the FamilieSCN2A Foundation. FamilieSCN2A hopes to accelerate the development of therapeutic treatments and disease-modifying advancements for those living with changes in the SCN2A gene.

**SCN2A Clinical Trial Readiness Study**

To determine the performance of specific functional, adaptive behavior, and related measures over time in children with SCN2A-DEE. This study will provide information on the stability of these measures over time and the extent to which they change with age. For therapeutic trials in which these measures could be used as primary or secondary outcomes, the data from this study will provide the basis for designing an efficient and robust trial.
Contract Research Agreements

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 and 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.

SCN2A Global Scientific Collaboration Calls

To facilitate open collaboration amongst the global scientific community, we host quarterly calls. These 90 minutes calls are well attended and the scientists take turns presenting their pre-published work and moderating a robust discussion where everyone shares learnings. Many collaborative projects have stemmed form these calls which expedites overall progress and reduces duplication.
The FamilieSCN2A Foundation raises public awareness of SCN2A related disorders through various platforms and media in addition to establishing an international presence of the disorder through awareness campaigns and initiatives.

Our awareness initiatives include:

- **International SCN2A Awareness Day**: Held annually on February 24th, FamiliesSCN2A engages families across the globe for International SCN2A Awareness Day.

- **SCN2A Warrior Wednesday**: Weekly on Wednesdays, FamilieSCN2A highlights a child living with SCN2A throughout our social media.

- **HOPE**: FamilieSCN2A disseminates a quarterly newsletters to constituents with regular research, awareness, and event updates.

- **Social Media Presence**: We have an active presence on social media. Join us on Facebook, Twitter, Instagram, Pintrest and YouTube.
We are dedicated to providing support to individuals living with SCN2A related disorders and their families. We are proud to offer the following mechanisms to help those affected by SCN2A:

**SUPPORT PROGRAMS INCLUDE:**

**NEWLY DIAGNOSED TOOLKIT**

**BIRTHDAY CLUB**

**GLOBAL SUPPORT NETWORK**

**FAMILY CONFERENCE & SCHOLARSHIP PROGRAM**
We are dedicated to providing support to individuals living with SCN2A related disorders and their families. We are proud to offer the following mechanisms to help those affected by SCN2A related disorders.

SUPPORT PROGRAMS INCLUDE:

- **SCN2A FAMILY EVENT GRANT**
- **COVID-19 FINANCIAL RELIEF GRANT**
- **PATIENT ASSISTANCE GRANT**
The FamilieSCN2A Foundation advocates for patients with SCN2A related disorders to improve early diagnosis, and empower families and professionals with education that directly relates to better care. We join forces with other rare diseases to effect change in policy and access to treatments.

ADVOCACY PROGRAMS INCLUDE:

RX: HOPE VIRTUAL SERIES

PARTNERSHIPS & COLLABORATION

BIENNIAL SCN2A FAMILY & PROFESSIONAL CONFERENCE
1. To coordinate and collaborate with researchers around the world to find the cause of and cure for SCN2A related disorders.

2. To increase public awareness about the severity and prevalence of SCN2A related disorders.

3. To provide educational and emotional support for children and families suffering from SCN2A related disorders.
Coordinate and collaborate with researchers around the world to find the cause of and cure for SCN2A-related disorders

OBJECTIVE #

1.1 Create and support an international Clinical Trial Readiness Study for SCN2A patients

1.2 Facilitate regular communication between researchers and clinicians to encourage increased knowledge of SCN2A, standard of care and possible treatments/cures

1.3 Grow the FamilieSCN2A Action Potential Grant Program to encourage scientific investigation into SCN2A
To increase public awareness about the severity and prevalence of SCN2A related disorders

OBJECTIVE #

2.1 Create clinician information and reference guide to aid clinicians in standard of care

2.2 Support early detection and genetic testing protocol research with a special focus on underserved populations

2.3 Increase public awareness of SCN2A through various mediums; create and sustain a strategic communications plan

2.4 Participate and promote awareness at family and/or professional centered events and conferences
HOW WE'LL GET THERE

Provide educational and emotional support for children and families suffering from SCN2A related disorders

OBJECTIVE #

3.1 Establish network of SCN2A coordinated care referral centers (Centers of Excellence)

3.2 Facilitate SCN2A Community Grant Programs to fund and promote regional family connections

3.3 Provide New Family Outreach to all recently diagnosed families in the SCN2A Community
OUR 2021 - 2022 GOALS

- Complete recruitment for Clinical Trial Readiness Study (CTRS)
- Form a corporate advisory committee for CTRS
- Fund mouse models for each of the SCN2A phenotypes
- Award 4 Action Potential Grants for post-doc research
- Take compound to clinical trial at Nationwide Children's Hospital
- Increase direct patient support through various programs
- Host FDA Listening Session and apply for a PFDD
- Recruitment and talent management for BOD
- Create system for customer and donor relations
- Increase social media and reputation outreach