2021 VISION MISSION VOICE

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

Opening April 1st!

SCN2A
CTRS
CLINICAL TRIAL READINESS STUDY

To stay informed, please make sure we have your current contact information. [Click here]() or email research@scn2a.org.

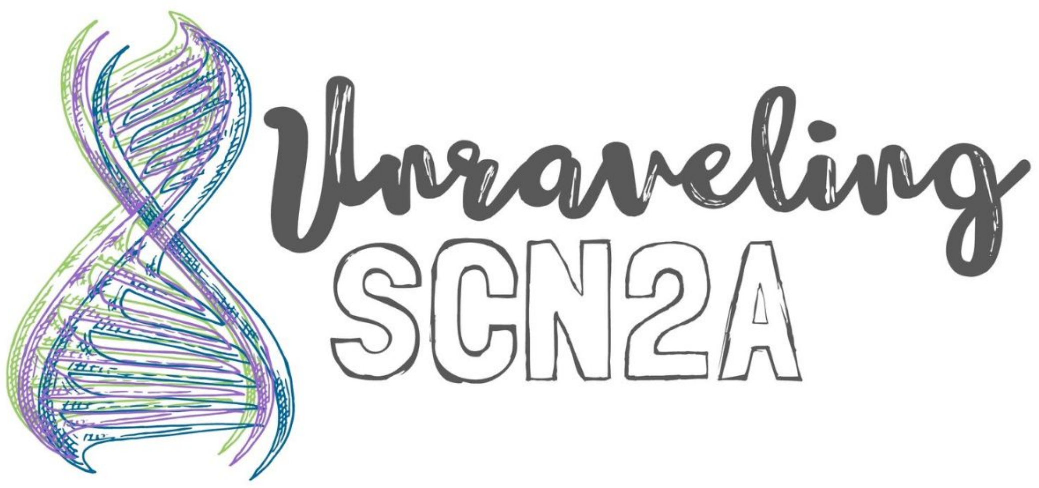
Ciitizen is a company using technology and machine learning to supplement the current Natural History Study model by digitally collecting and analyzing critical patient data in order to scale rare disease research without exponential cost. Most importantly, this model allows patients direct access to their consolidated, digitized data that also uniquely summarizes their experience, which can be used to get second opinions or share with multiple

providers to facilitate and improve their care.
Sign up takes 10 minutes. https://www.ciitizen.com/scn2a/

Watch: Ciitizen/SCN2A webinar
Thursday 3/11 at 2:00 pm ET
Click here to register for webinar

Let your voices be heard! What would you like to see happen regarding our **2021 Family and Professional Conference**? Please fill out this brief survey so that we can start making plans to serve you best. Thank you!

We are excited to introduce the first entry in our new blog:



FINDING YOUR VOICE

By: Michelle Lewis, FamilieSCN2A Foundation Vice President

"I raise up my voice—not so I can shout, but so that those without a voice can be heard" Malala Yousafzai

FINDING MY VOICE

On a cold January morning during the first week of what, unbeknownst to me, would become a 5 month long stay at our local children's hospital NICU, I was holding my daughter when I noticed every few minutes she would repeat this strange pattern with her body and face. I asked the nurse what she thought and she said to bring it up to the doctors during daily rounds. This would be the first time I would use my voice to advocate for my daughter.

Years later, I am still using my voice, like most SCN2A parents and caregivers, to advocate for my child, whether that be fighting to get better medical care, necessary therapies and services, appropriate access to education or essential equipment.

In the rare disease world, not only finding your voice but amplifying it loud enough so that others can hear it is a daunting task that many families simply don't have the energy or bandwidth to do. A few easy ways to make your voice heard include:

- **Share awareness posts from the** FamilieSCN2A Foundation's **public Facebook page with your friends and family.**
- **Share your** SCN2A story **on the FamilieSCN2A website.**
- **Apply for an** SCN2A Family Event Grant **and get local SCN2A families together.**

Maybe you have time to do these things, maybe you don't. If you ever get to a point where you can and want to do more, please consider volunteering your time or skills to helping the FamilieSCN2A Foundation amplify the community voice.

AMPLIFYING THE COMMUNITY VOICE

The FamilieSCN2A Foundation is dedicating our sixth year of existence to amplifying the voices of our community. Knowing that new treatments are on the horizon for the SCN2A community our goal is to create a unified

voice so that when the time comes, we are ready to advance treatments and cures.

One way we are amplifying our community voice is by participating in a Listening Session for Rare Diseases with the Food and Drug Administration (FDA). Listening sessions are an opportunity for patients and caregivers to share directly with FDA staff our experiences living with and managing SCN2A Disorders in our daily lives. Listening sessions provide the FDA with personal and clinical understanding of diseases and conditions, provide a common understanding of the most urgent needs, and inform drug development programs directly from OUR perspective. This session is one important way we can help inform regulatory decision-making when the time comes for new treatments and cures to be reviewed. The FamilieSCN2A Foundation is participating in a listening session with the FDA in April, 2021.

Another way we are working to create a unified voice for the SCN2A community is through the initiation of the SCN2A Clinical Trial Readiness Study (CTRS). Successful clinical trials for a drug or biologic, like gene therapy, must demonstrate to the FDA that it is not only safe and effective but that the outcomes, or endpoints, of the treatment are measurable and desirable in the patients. When designing a clinical trial, it is critical to use appropriate assessment tools to accurately measure and demonstrate even small changes in a patients' abilities. A measurement should be able to show a range of abilities and at different ages as ability levels change. Unfortunately, many validated measures do not accurately assess the spectrum of SCN2A Disorders. The goal of the CTRS is to determine the performance of various assessment tools over time for SCN2A. The data from this study will provide the basis for designing efficient and robust clinical trials.

Setting these measurement tools up in advance with the CTRS will ultimately help save time and resources. There will be a data use agreement as part of the patient's informed consent so that industry groups can have access to de-identified data. This is the kind of collaboration that will expedite the path to cures for SCN2A related disorders.

A CRITICAL NOTE ABOUT DATA

One of the most impactful ways you can make your voice heard is by participating in data collection. For the past three years, we have worked tirelessly to make data collection as simple and non-invasive as possible for our already stressed and exhausted families, with the ultimate goal of having all SCN2A data collected in one place, where not only researchers and industry groups have access to it, but so do families. While this was the ultimate goal, we hit a few roadblocks along the way which has led to SCN2A data being collected in a few different repositories. We are continuing to push all groups that are collecting data to make it open and accessible to any and all validated researchers and groups attempting to advance understanding, treatments and cures for SCN2A. We know how exhausting living with SCN2A is for families and we will continue to advocate and amplify your voice so that one day we will achieve our vision of a world without SCN2A-related disorders.

Current SCN2A Data Repositories:

- SCN2A CTRS **(enrollment begins April 1)**
- Simons Searchlight
- Ciitizen Electronic Medical Records Collection
- SCN2A Natural History Study

The FamilieSCN2A Foundation would love to hear how you have used your voice to advocate for your loved one impacted by SCN2A Disorders. Please send your story, along with a recent picture, to info@scn2a.org. Sharing stories of how we use our voices helps to make us heard! We would love to profile your story on our social media platforms.

Look for the 'Unraveling SCN2A' blog on our website soon!

View this email in your browser