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

Spring 2022

Dear Friend,

This year we are celebrating our 7th anniversary of becoming a nonprofit! Seven years of helping families and funding life-saving research. We are writing to share the “audacious optimism” we at The FamilieSCN2A Foundation feel about building our future thanks to our generous supporters.

Changes in the SCN2A gene are recognized as one of the most common single-gene causes of autism and early onset epilepsy. Understanding the cause of these devastating disorders has inspired researchers to find new treatments, which are on the horizon.

Over the last 7 years, we have funded nearly $1,000,000 in research to find a cure for SCN2A-related disorders. FamilieSCN2A supports research for treatments in the areas of drug repurposing, new compounds, and small molecules, as well as gene altering therapeutics. We firmly believe that through our continued dedicated research funding we will not only find a cure for SCN2A-related disorders, but also contribute to the larger autism and epilepsy communities.

In addition to research, we offer a Patient Assistance Grant program, which sets aside funds to help SCN2A patients with necessary equipment and therapy that are not covered through other means. For the last 2years, we helped to meet the needs of many families devastated by loss of income due to the pandemic by offering Covid-19 Emergency Grants. We are proud to say we were able to fund nearly every grant application.

In honor of our 7th anniversary, please consider making a thoughtful donation to support the work of the Foundation as we continue to help children and families with SCN2A-related disorders fight through this disease.

Any size donation truly does make an impact. If you are able to donate $5, $50, $500 or $5,000 today, you are moving our mission forward through research and directly helping families who struggle daily through this journey. Please click on the Donate button below or visit scn2a.org for more information.

Gratefully,

Jennifer Burke, President

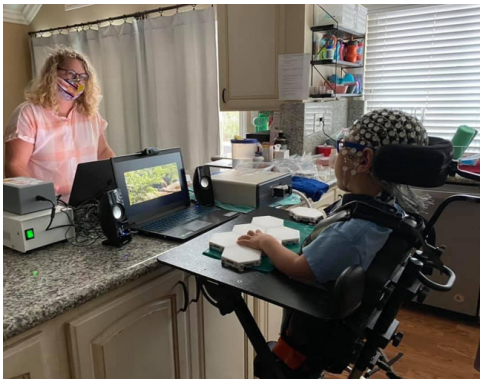
Leah Myers, Founder & Executive Director

Donate

FAMILIESCN2A® annual campaign highlights

CLINICAL TRIAL READINESS STUDY

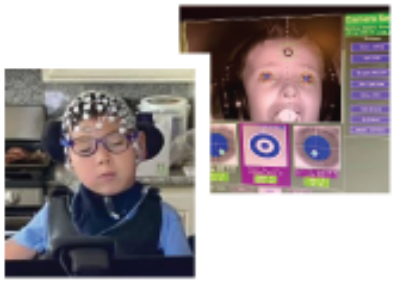
Preliminary results of the first ever SCN2A Clinical Trial Readiness Study are available now at www.scn2a.org/research.html, with the full report coming later this year. This study will provide meaningful endpoints for our industry partners to target when designing clinical trials and will help in the FDA approval process for new treatments. To further prepare for clinical trials, we held an FDA Listening Session to educate regulators on real world patient experiences with SCN2A-related disorders.



RESEARCH

The Foundation-sponsored project at Nationwide Children's Hospital in Ohio is completing a safety study for the molecule CuATSM, which is currently in clinical trials for other neurodegenerative disorders and has years of positive safety data in adults. Efficacy has been shown in SCN2A patient cell lines and mice. If the safety data is positive and the FDA agrees, we will possibly begin clinical trials in SCN2A patients this year!

In addition, there are two other potential treatments entering trials in Q2 and while advancements in gene therapy are happening at lightning speed in many diseases, they may still be a couple of years away for SCN2A.



Grant recipient, Dr. Caitlin Hudac, took her SCN2A research on the road in 2021

GRANT RECIPIENTS OF THE CHAN ZUCKERBURG INITIATIVE

"We are from Venezuela and 2 months ago we moved from Chile to the US. During the initial days we learned that the system to get access to a doctor…was totally different and we are running out of all medication for epilepsy treatment…I called

the Foundation and in two days it was resolved. We got an appointment with an amazing team and are able to continue the treatment that has controlled the seizures of my daughter. Sometimes it is lucky, but this was 90% support from the FamilieSCN2A Global Support Network and the beautiful people on it. One of the parents even offers us her appointment she has for her beautiful daughter to us...this is amazing, and I can't find a bigger word in English to say THANK YOU to this team...The Foundation helped us to resolve a problem but also gave us energy and hope."

FamilieSCN2A is proud to participate in the Chan Zuckerberg Initiative's Rare As One project. As a grant recipient we are poised to build our organizational capacity, community reach and research network over the next three years. (The grant does not fund research—that is up to us, with your support.)

SCN2A AWARENESS DAY

"By raising the awareness of this devastating disorder, we hope to continue SCN2A's efforts to capture the interest of the scientific community and pharmaceutical companies to discover new treatments not only to save the lives of patients, but also to help alleviate the tremendous burden on families."

The significance of the February 24 (2/24) date comes from the location of the gene on the long (q) arm of chromosome 2 at position 24.3. The gene SCN2A encodes instructions to make a protein in the brain called a sodium channel which plays a key role in a cell's ability to generate and transmit electrical signals. Pathogenic variants that affect the SCN2A sodium channel impair the flow of sodium ions in the brain. When there is a deletion or mutation of this gene it has been identified to cause autism, epilepsy and other neurological issues such as movement disorders, dystonia and dysautonomia.