

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.

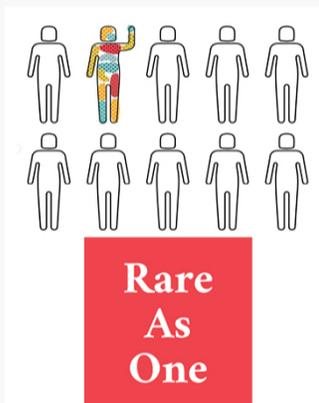


Grant recipient, Dr. Caitlin Hudac, took her SCN2A research on the road in 2021

ongoing family support

"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."

GRANT RECIPIENTS OF THE CHAN ZUCKERBURG INITIATIVE



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.



BUILDING
TOWARDS
A CURE

2022

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