What is the FamilieSCN2A Foundation?

The FamilieSCN2A Foundation is an organization created by parents of children diagnosed with **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 disorders. Our **mission** is to improve the lives of those affected by SCN2A disorder through clinical research, effective treatments, public awareness, early detection, patient advocacy, and family support.

We are a registered 501(c)(3) organization run entirely by parent volunteers.

Email us: info@scn2a.org

P.O. Box 82, East Longmeadow, MA 01028

www.scn2a.org

CONNECT WITH US!

International SCN2A Awareness Day: February 24

Celebrate International SCN2A Awareness Day on February 24th!

Significance Of The Date 2/24:
Located on the long (q) arm of chromosome **2** at position **24**.3 (2/24) the SCN2A is sodium channel, voltage gated, type II alpha subunit. Sodium ion channels are proteins in cells that allow sodium to pass to the inside. Sodium ion channels play a key role in a cell's ability to generate and transmit electrical signals.

SCN2a Awareness Day will help us accomplish the FamilieSCN2A Foundation's mission.

Visit: **ww.scn2a.org** to find out more.

INTERNATIONAL SCN2A AWARENESS DAY
February 24th

Visit our web site to find out how you can contribute toward helping those diagnosed with SCN2A. Let's find a cure!

SCN2A RELATED AUTISM & EPILEPSY
FAMILIESCN2A FOUNDATION

SCN2A

A sodium ion channel gene

What is SCN2A?

SCN2A is a sodium ion channel gene located on chromosome 2.
Sodium ion channels are proteins in cells that allow sodium to pass to the inside. Sodium ion channels play a key role in a cell's ability to generate and transmit electrical signals.

Seizure Disorders Associated With Changes In The SCN2A Gene:

Epilepsy, Benign Familial Infantile Seizures, Early Infantile Epileptic Encephalopathy, Ohtahara & West Syndrome, Generalized Epilepsy with Febrile Seizures, Migrating Partial Epilepsy of Infancy (MPEI), Infantile Spasms

Other Medical Challenges Associated With SCN2A:

ASD, ADHD, Global & Speech Delays, Intellectual Disability, Movement Disorders Including Ataxia & Dystonia, GI, GERD and Feeding Issues, Cortical Visional Impairment, Sleep Issues, Urology problems, Neuropathic Pain, Autonomic Dysfunction, and Cerebral Palsy

*Some kids are labeled as CP for insurance purposes

Newly Diagnosed?

Currently, there is not a single clinical presentation or phenotype for SCN2A. Researchers are actively studying variants of this gene. The most important step you can take is registering your child through the Simons VIP database where researchers collect data for their studies. Visit: **www.scn2a.org**

The FamilieSCN2A Foundation has partnered with Simons VIP because of their desire to study genetic changes of SCN2A that cause both Autism and Epilepsy.

You can connect with other families affected by SCN2A throughout the world by finding us on Facebook and requesting to join our **PRIVATE** group. You can find us by searching for: **FamilieSCN2A Community Discussion Group.**

SCN2A and Autism

It has been discovered that some mutations in SCN2A appear to dampen brain activity and are linked to autism; others have the opposite effect and may lead to seizures during infancy. (Ben-Shalom R. *et al. Biol. Psychiatry,* 2017)

SCN2A codes for a channel that allows sodium ions to traverse neurons. In the past two years, it has emerged as one of the genes mostly strongly linked to autism. (Jessica Wright, spectrumnews.org)

Common Specialists Seen By Children With SCN2A Include:

Neurologist, Neuropsychologist, Endocrinologist, Orthopedist, Gastroenterologist, Urologist, PM&R/ Physiatrist, Palliative, Complex Care, Sleep Specialist, Ophthalmologist, Autism Specialist, Occupational, Speech, & Physical Therapists

Children with SCN2A will benefit from a team approach with multiple specialists involved in their care.