



## When is International SCN2A Awareness Day?

**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 gene encodes the voltage-gated sodium channel Nav1.2 mainly located in the brain. Sodium ion channels are proteins in cells that allow sodium to enter inside to generate and transmit electrical signals.

Visit: [www.scn2a.org](http://www.scn2a.org) to find more information and how to help those diagnosed with SCN2A Disorders.  
Let's find a cure!

Epilepsy

Autism

Cerebral Palsy



# SCN2A

## Who is the FamilieSCN2A Foundation?

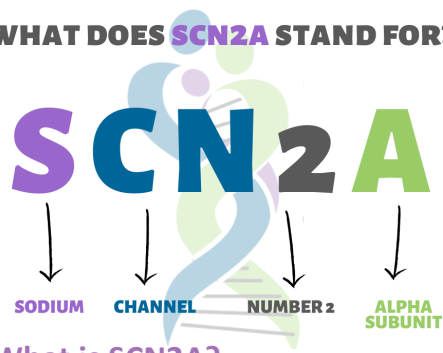
The FamilieSCN2A Foundation is a registered 501c3 non profit organization created by parents of children diagnosed with **Epilepsy** and **Autism** as a result of a change in the SCN2A gene.

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 Disorders through clinical research, effective treatments, public awareness, early detection, patient advocacy, and family support.

Follow us on Facebook (SCN2A Related Autism & Epilepsy: The FamilieSCN2A Foundation), Twitter (@FamilieSCN2A), and Instagram (#FamilieSCN2AFoundation).



## WHAT DOES SCN2A STAND FOR?



## What is SCN2A?

SCN2A is a sodium ion channel gene located on chromosome 2. It encodes the alpha subunit of the voltage-gated sodium channels (Nav1.2) mainly located in the brain. These channels play an essential role in a cell's ability to generate and transmit electrical signals. A change in the gene can alter the function of the channel and affect the way neuronal impulses are conducted.

## Associated Medical Conditions

- Autism Spectrum Disorder
- Autonomic Dysfunction
- Cerebral Palsy (spasticity, hypotonia)
- Cortical Vision Impairment
- Epilepsy
- GI Dysfunction (Reflux & constipation)
- Intellectual Disability
- Movement Disorders (chorea, ataxia, dystonia)
- Neuropathic Pain
- Sleep Disorders
- Speech and Language Deficit
- Urology problems (infections & urinary Retention)

## How Does SCN2A Present?

The current published literature suggests two main presentations: gain of function of the sodium channel or a loss of function of the sodium channel. A gain of function mutation leads to improper channel closing, causing more sodium to enter the cells and excess neuronal firing or excitability. A loss of function mutation leads to improper channel opening, causing less sodium to enter the cells and insufficient neuronal firing. Having a good understanding of this can help guide proper therapeutic decisions.

### SCN2A DISORDERS

INFANTILE EPILEPTIC ENCEPHALOPATHY	BENIGN (FAMILIAL) INFANTILE SEIZURES	AUTISM / INTELLECTUAL DISABILITY
INFANTILE-ONSET SEIZURES, FOLLOWED BY NEURODEVELOPMENTAL DELAY	CHARACTERIZED BY INFANTILE-ONSET SEIZURES THAT RESOLVE BY AGE 2	GLOBAL DEVELOPMENTAL DELAY IN SOCIAL AND LANGUAGE MILESTONES; WITH OR WITHOUT SEIZURES
INCREASED	NORMAL	REDUCED
INFANTILE EPILEPTIC ENCEPHALOPATHY	BENIGN (FAMILIAL) INFANTILE SEIZURES	AUTISM / INTELLECTUAL DISABILITY

## Associated Epilepsy Syndromes

- Benign Familial Infantile Seizures
- Early Infantile Epileptic Encephalopathy (e.g. Ohtahara & West Syndrome)
- Later onset epilepsy with ASD
- Lennox-Gastaut Syndrome
- Migrating Partial Epilepsy of Infancy

## Newly Diagnosed?

Knowledge is power. Our website is full of resources for families and professionals wanting to learn more about SCN2A. We have also partnered with Simons Searchlight to collect data for researchers studying this disease. Visit [www.scn2a.org](http://www.scn2a.org)

## Common Specialists Seen By Patients With SCN2A

- Complex Care
- Developmental Pediatrician
- Endocrinologist
- Gastroenterologist
- Geneticist
- Neurologist
- Neuropsychologist
- Ophthalmologist
- Orthopedist
- Palliative Care
- Psychiatrist
- Pulmonologist
- Urologist
- Occupational, Physical, Speech and Vision Therapist

*Patients with SCN2A benefit from a multi-disciplinary team approach with numerous specialists involved to deliver comprehensive care.*