

# INTERNATIONAL SCN2A DAY

FEBRUARY 24TH

2q24.3



Celebrate International SCN2A Awareness Day on February 24th!

February 24th (2/24) was chosen as International SCN2A Awareness Day because SCN2A is located on the long (q) arm of chromosome 2 at position 24.3.

## STAY IN THE FAMILY

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 @FAMILIESCN2A FOUNDATION

SCN2A-RELATED AUTISM & EPILEPSY  
**FAMILIESCN2A**<sup>®</sup>  
FOUNDATION  
WWW.SCN2A.ORG



P.O. BOX 4260 GETTYSBURG, PA 17325



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## WHO WE ARE

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

## OUR VISION

A world with effective treatments and cures for all SCN2A-related disorders

## OUR MISSION

To accelerate research, build community and advocate to improve the lives of those affected by SCN2A-related disorders around the world.



## COMMON SPECIALISTS SEEN BY PATIENTS WITH SCN2A-RELATED DISORDERS

- Complex Care
- Developmental Pediatrician
- Endocrinologist (Bone Health)
- Epileptologist
- Gastroenterologist
- Geneticist
- Neurologist
- Neuropsychologist
- Ophthalmologist
- Orthopedist
- Palliative Care
- Psychiatrist
- Pulmonologist
- Urologist
- Occupational, Physical, Speech & Vision Therapists

# SCN2A



## WHAT IS SCN2A?

SCN2A is a sodium channel gene located on chromosome 2. It encodes the alpha subunit of the voltage-gated sodium channels (Nav1.2), found primarily 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

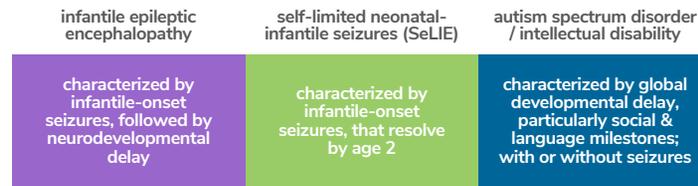
- Autism Spectrum Disorder
- Autonomic Dysregulation
- Cortical Visual Impairment
- Epilepsy
- GI Dysfunction
- Intellectual Disability
- Motor Delay
- Movement Disorders
- Neuropathic Pain
- Orthopedic Problems
- Sleep Disorders
- Speech & Language Deficits
- Urologic Disorders

## HOW DO SCN2A-RELATED DISORDERS PRESENT

The published literature suggests two main ways the gene can be disrupted: Gain of Function (GoF) or Loss of Function (LoF) of the sodium channel. Recently, scientists have discovered many variants that fall in the middle of the scale and have more of a mix or change of function.

A GoF variant makes the channel more excitable, typically leading to early infantile-onset seizures.

A LoF variant reduces the excitability or destroys channel function altogether, typically leading to autism spectrum disorder and/or intellectual disability. Approximately 30% of these patients may develop later onset epilepsy.



## ASSOCIATED EPILEPSY SYNDROMES

- Benign Familial Infantile Seizures
- Early Infantile Epileptic Encephalopathy
- Ohtahara Syndrome
- West Syndrome / Infantile Spasms
- Lennox-Gastaut Syndrome
- Migrating Partial Epilepsy of Infancy
- Epileptic Encephalopathy with Continuous Spikes-and-Waves during Sleep (EE-CSWS)

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

## NEWLY DIAGNOSED?

**Knowledge is power.** Our website is full of resources for families and professionals wanting to learn more about SCN2A.

Discover the latest research on SCN2A and join our Global Family Network, a community where affected families can turn for support and guidance from others in similar circumstances. **You are not alone.**



[WWW.SCN2A.ORG](http://WWW.SCN2A.ORG)