



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.

FOLLOW US ON SOCIAL MEDIA



@FAMILIESCN2A #SCN2A #CURESCN2A



@FAMILIESCN2AFUNDATION



FAMILIESCN2AFUNDATION



FAMILIESCN2A FOUNDATION



FAMILIESCN2A



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.

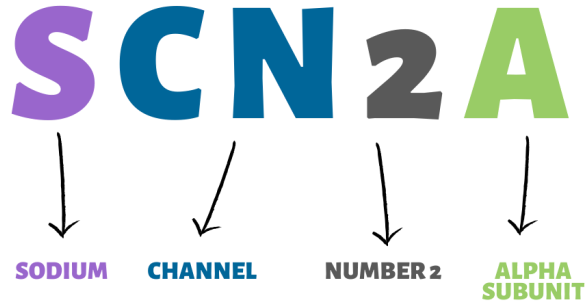
OUR VISION

To find effective treatments and a cure for SCN2A disorders.

OUR MISSION

To improve the lives of those affected by SCN2A disorders through research, public awareness, family support, and patient advocacy.

WHAT DOES SCN2A STAND FOR?



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
- Sleep Disorders
- Speech and Language Deficits
- Urologic Disorders

HOW DOES SCN2A PRESENT?

The current published literature suggests two main presentations: Gain of Function of the sodium channel or Loss of Function of the sodium channel.

A Gain of Function variant makes the channel more excitable, typically leading to infantile-onset seizures.

A Loss of Function variant reduces the excitability or destroys channel function altogether, typically leading to autism spectrum disorder and/or intellectual disability.

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.**

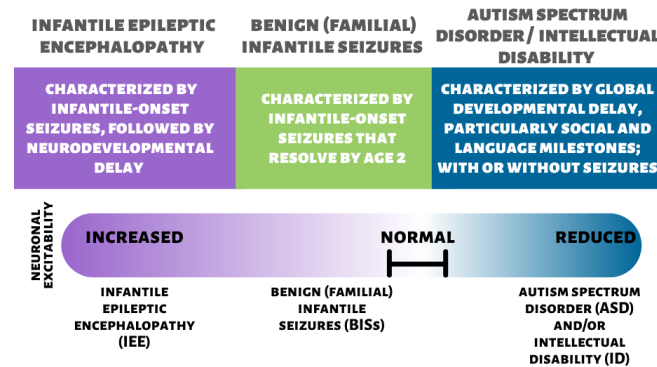
WWW.SCN2A.ORG

COMMON SPECIALISTS SEEN BY PATIENTS WITH SCN2A

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

PATIENTS WITH SCN2A BENEFIT FROM A MULTI-DISCIPLINARY TEAM APPROACH WITH NUMEROUS SPECIALISTS INVOLVED TO DELIVER COMPREHENSIVE CARE.

SCN2A DISORDERS



ASSOCIATED EPILEPSY SYNDROMES

- Benign Familial Infantile Seizures
- Early Infantile Epileptic Encephalopathy
- Later Onset Epilepsy with ASD
- Lennox-Gastaut Syndrome
- Migrating Partial Epilepsy of Infancy