



FAMILIESCN2A FOUNDATION

WELCOME TO OUR COMMUNITY

A GUIDE FOR FAMILIES NAVIGATING AN SCN2A DIAGNOSIS



"FAMILIES" IS PART OF OUR NAME FOR A REASON. SCN2A-RELATED DISORDERS AFFECT THE ENTIRE FAMILY. OUR TEAM OF LEADERS STRIVE EVERY DAY AND IN EVERY WAY TO IMPROVE THE LIVES OF NOT ONLY THE PATIENT, BUT THE ENTIRE FAMILY.



WELCOME TO

The FamilieSCN2A Foundation

If you're holding this booklet, you may be feeling overwhelmed, uncertain, or full of questions. An SCN2A-related disorder (SRD) diagnosis can change everything in an instant, and it's okay if you don't know where to begin.

Most importantly: you are not alone.

SRDs affect each child and family differently. There isn't one "right" path or timeline. Whatever you're feeling—grief, fear, relief at finally having answers, or a mix of it all—is valid.

This guide is here to support your first steps, at your own pace. You don't need to read it all at once, and you don't need to understand everything today. Think of it as something you can come back to when questions come up, appointments feel heavy, or you simply need reassurance that others have walked this road too.

Inside, you'll find a plain-language overview of SCN2A, questions to bring to your care team, information on therapies and treatment options, and ways to connect with research and community when you're ready. Nothing here is meant to rush you, only to support you.

At the FamilieSCN2A Foundation, families are at the heart of everything we do. Our community spans the globe, connected by shared experience, compassion, and the belief that no family should face this diagnosis alone. Whether you reach out today or later on, we'll be here.

Take a deep breath. Take it one step at a time. You've found a community that understands.

With care and hope,
The FamilieSCN2A Foundation Team

WHO WE ARE

The FamilieSCN2A Foundation

The FamilieSCN2A Foundation is a global nonprofit organization dedicated to supporting individuals and families affected by SCN2A-related disorders. Founded by families and guided by lived experience, we understand how complex and overwhelming this diagnosis can be. We are here to ensure that no family has to navigate this journey alone.

Our mission is to accelerate research, build community, and advocate to improve the lives of those affected by SCN2A-related disorders around the world. Through collaboration with families, clinicians, and researchers, we work to advance understanding, support meaningful connections, and move closer to effective treatments and cures.

We are a registered 501(c)(3) organization.

SCN2A RELATED DISORDERS (SRDs)

What does SCN2A mean?

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 nerve impulses are conducted.

ASSOCIATED MEDICAL CONDITIONS

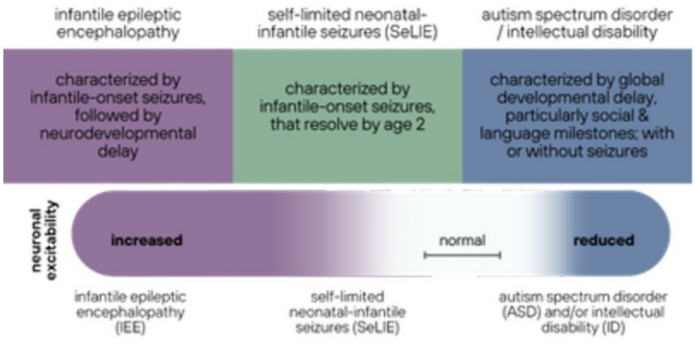
- Epilepsy/Seizure Disorders
- Movement Disorders
- Autonomic Dysfunction
- Stomach/Bowel problems
- Urinary issues
- Feeding issues
- Neuropathic pain
- Muscle tone issues (Hypotonia/Hypertonia)
- Autism Spectrum Disorder
- Attention Deficit Hyperactivity Disorder
- Global Developmental Delays
- Intellectual Disability
- Speech Disorders
- Cortical Visual Impairment
- Sleep issues

Changes in SCN2A are among the most common causes of neurodevelopmental disorders. Even in people with the same variants, clinical presentation may vary.

How do SRDs Present?

SCN2A changes can affect people in different ways. These changes are on a spectrum from mild to severe effects. Some SCN2A changes are called gain-of-function (GoF). These make the sodium channel in the brain work too much. This usually causes seizures right after birth and can lead to more serious brain development problems.

Other changes are called loss-of-function (LoF). These make the channel work less or not at all. This type is more often linked to developmental delays, autism, intellectual disability, and sometimes seizures that start in early childhood. There are also mixed-function changes, where the effects are a mix of both types. These often start with a kind of seizure called infantile spasms in late infancy and can also involve movement problems, autism, and severe epilepsy.



Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., Sanlnocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. https://doi.org/10.1017/9781009530361

SCN2A-RELATED EPILEPSY

ASSOCIATED EPILEPSY SYNDROMES

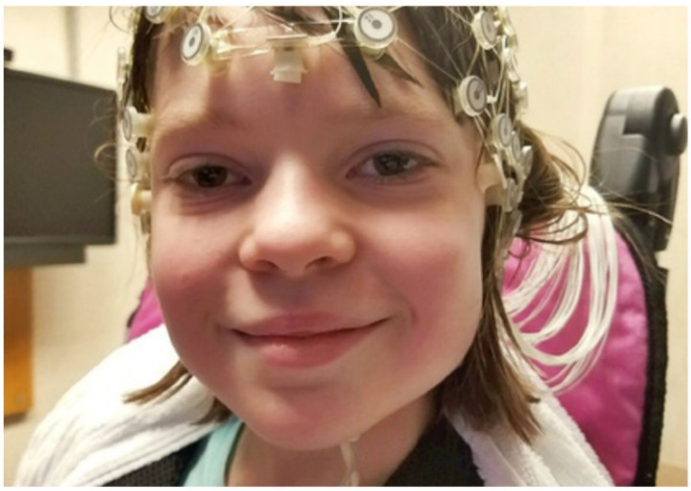
- Self Limited Infantile Epilepsies (SeLIE)
- Early Infantile Epileptic Encephalopathy (type 11)
- Ohtahara Syndrome
- Infantile Epileptic Spasms Syndrome
- Lennox-Gastaut Syndrome (LGS)
- Generalized Epilepsy with Febrile Seizures
- Epilepsy of Infancy with Migrating Focal Seizures
- Later-onset epilepsy with ASD

Seizures in SCN2A-related disorders vary and may change over time. Sudden unexpected death in epilepsy (SUDEP) is the sudden, unexplained death of someone with epilepsy. While risk is not elevated for all individuals with SCN2A, it is highest in those with early-onset, severe, or refractory epilepsy, particularly when convulsive seizures are present.

How Rare are SRDs?

SRDs affect an estimated 11 out of every 100,000 births, resulting in more than 400 children born each year in the United States. While loss-of-function (LoF) variants are predicted to outnumber gain-of-function (GoF) variants by approximately 7 to 1, GoF variants are often associated with earlier onset and more severe clinical presentations.

Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., SanInocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. https://doi.org/10.1017/9781009530361



GAIN OF FUNCTION IN SCN2A
& SODIUM CHANNEL BLOCKERS (SCB)

- There is a correlation between age at disease onset, response to SCBs and the functional properties of variants in children with SCN2A-related epilepsy.
- Variants associated with early infantile epilepsy tend to result in increased sodium channel activity with gain-of-function.
- SCBs were often associated with clinically relevant seizure reduction or seizure freedom in children with early infantile epilepsies (<3 months), whereas other anti-epileptic drugs were less effective.
- Clinical Trials are available for this subtype. Please visit SCN2A.ORG for the latest therapeutic pipeline.

Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., SanInocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. https://doi.org/10.1017/9781009530361

EXAMPLES OF SODIUM CHANNEL BLOCKERS

phenytoin, carbamazepine, oxcarbamazepine, lacosamide, lamotrigine, zonisamide

**names my vary depending on country*

SCN2A-RELATED AUTISM

How Is SCN2A Related Autism Different?

SCN2A-related autism is caused by a specific gene change that disrupts how brain cells communicate and is often associated with multiple co-occurring medical and developmental conditions. Individuals with SCN2A-related autism tend to have challenges in these specific areas:

- Social interaction, Communication and sensory processing.
- Restricted, repetitive, or stereotyped patterns of behavior
- Motor planning difficulties
- Gastrointestinal issues

SCN2A related autism can present both with and without epilepsy.

It is estimated that one-third of SCN2A autism patients will develop epilepsy. Sodium channel blockers were rarely effective in later onset epilepsy typically seen in the SCN2A autism population.

Source: Wolff, et al. (2017). Genetic and phenotypic heterogeneity suggest therapeutic implications in SCN2A-related disorders. Brain, 140(5):1316-1336

How Rare is SCN2A-Related Autism?

Changes in the SCN2A gene are among the leading single-gene causes of autism. Overall, SCN2A-related disorders affect an estimated 11 out of every 100,000 births, resulting in more than 400 children born each year in the United States with an SCN2A-related disorder. Among these, loss-of-function (LoF) variants are predicted to outnumber gain-of-function (GoF) variants by approximately 7 to 1.

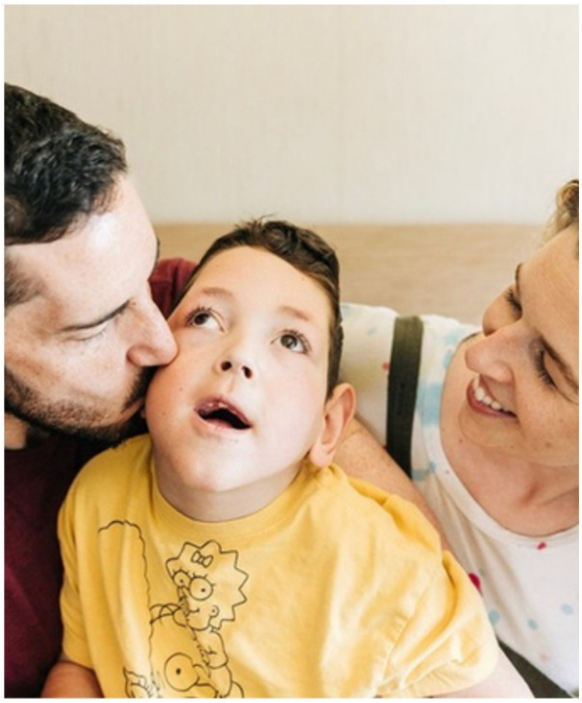
Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., SanInocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. https://doi.org/10.1017/9781009530361

LOSS OF FUNCTION SCN2A

- In contrast to gain of function variants that contribute to seizures, ASD-associated SCN2A variants dampen or eliminate channel function.
- ASD-associated variants affect the electrical properties of NaV1.2 channels by reducing the function of the sodium channel.
- Loss-of-function can range from stopping the channel from being made to blocking the pore through which sodium needs to flow for the channel to function.
- There is a clear correlation between loss of function variants and ASD.

Source: George, Jr, A. L., Abbott, M., Bender, K. J., Brunklaus, A., Demarest, S., Egan, S., Haviland, I., Kearney, J. A., Myers, L. S., Olson, H. E., Sanders, S. J., SanInocencio, C., Symonds, J., & Thompson, C. H. (2024). SCN2A-Related Disorders. Elements in Genetics in Epilepsy. https://doi.org/10.1017/9781009530361

Autism is more commonly known as Autism Spectrum Disorder (ASD) because of the wide variation in the type and severity of symptoms that people experience. ASD is a developmental disability that is caused by differences in the way the brain functions.



WHAT'S NEXT?

Receiving an SCN2A diagnosis can bring clarity, but also lead to more questions. That's okay. Understanding your child's diagnosis and variant is an important step in navigating your child's care and advocating for their future.

You don't have to figure it all out alone, which is why the FamilieSCN2A Foundation has many resources for you and your family to learn more, get connected, and help move research forward.

UNDERSTANDING SCN2A

SCN2A-related disorders are severe, lifelong neurological conditions, and their impact can vary widely from person to person. There is no one-size-fits-all outcome. Each person's path is shaped by their specific genetic change, how the condition affects them, and the care and support they receive.

Individuals do not outgrow SRDs, but there is real hope. Earlier diagnosis, growing understanding, and more tailored treatment approaches are helping to improve care and outcomes for many individuals.

The following pages outline treatment and therapy options based on current international clinical experience and consensus, reviewed by the FamilieSCN2A Foundation's Medical and Scientific Advisory Board, to support informed conversations with your child's care team.

SCN2A TREATMENTS & THERAPIES

***Treatment decisions should always be made with your child's medical team.*

Anti Seizure Medications (ASM)

General considerations for ASMs in SCN2A-related disorders:

- Responses and side effects can vary and are often monitored over time.
- Seizure control may play a role in long-term outcomes.
- Medication effectiveness can differ based on an individual's SCN2A presentation (phenotype).

GAIN OF FUNCTION PHENOTYPE

- Sodium channel blockers (SCBs) are commonly used as first-line treatment. Families may hear about medications such as **phenytoin, lacosamide, oxcarbazepine, carbamazepine, lamotrigine, cenobamate eslicarbazepine and zonisamide.**
- Other medications have been explored in smaller studies or individual cases, including **cannabidiol, lidocaine, and topiramate.**

LOSS OF FUNCTION PHENOTYPE

- AVOID sodium channel blockers
- ASM to consider include:
  - **Benzodiazepines (Clobazam, Clonazepam), Levetiracetam, Valproate, Vigabatrin, Topiramate, Stiripentol, Rufinamide**
- Less mainstream medications (with limited data) to consider:
  - **Ethosuximide, Felbamate, Acetazolamide (for episodic ataxia)**

Additional Treatment Options

- Dietary Therapies: **Ketogenic Diet, Modified Atkins Diet, Low Glycemic Index Treatment**
- Neuromodulation: **Vagal Nerve Stimulator, Responsive Neurostimulation, Deep Brain Stimulation**
- Steroids and/or Vigabatrin: first line treatment for Infantile Spasms

To learn more about current research and the treatment pipeline, visit: https://www.scn2a.org/research/

Therapeutic Interventions

- Occupational Therapy
- Physical Therapy
- Vision Therapy
- Hippotherapy
- Hydrotherapy
- Music Therapy
- Applied Behavior Analysis (ABA)
- Sensory Integration Therapy
- Assistive Augmentative Communication (AAC) Therapy

DEVELOPMENTAL & EPILEPTIC ENCEPHALOPATHY

Developmental and Epileptic Encephalopathy (DEE) refers to conditions where an underlying genetic change leads to both severe epilepsy and developmental impairment. In DEE, development is affected by the genetic cause itself as well as by the ongoing impact of seizures and abnormal brain activity over time.

In SCN2A-related disorders, the term DEE applies to individuals—including those with both GoF and LoF variants—who have early-onset, often difficult-to-control seizures that significantly worsen developmental outcomes. While DEE is more frequently associated with early-infantile presentations, some individuals with SCN2A LoF variants who develop epilepsy also meet criteria for DEE when seizures and epileptiform activity are a major driver of developmental disability.

Not all people with SRDs have DEE; the term is used specifically when epilepsy itself meaningfully contributes to developmental impairment, beyond the effects of the genetic variant alone.

A clear DEE diagnosis helps guide the right therapies, anticipates co-occurring needs, and opens doors to specialized care, research studies, and clinical trials that can change outcomes.

WHERE TO **START**

- Subscribe to the FamilieSCN2A Foundation email list
- Explore the **SCN2A.ORG** website
- Register for SCN2A Town Hall meetings
- Watch our *"About SCN2A"* series on Youtube
- Download our *Family Research Checklist* →

CONTRIBUTE TO **RESEARCH**

If your family participates in data collection studies such as the **DRAGONFLY Study**, your data helps researchers:

- Determine which treatments are developed—and which are not
- Define the targets researchers and industry pursue
- Provide the evidence regulators and clinical trial designers require
- Accelerate therapies from lab to clinic
- Improve care and outcomes for families today
- Create a path forward for families who receive a new diagnosis

To learn more, visit:
www.scn2a.org/research/how-to-participate/

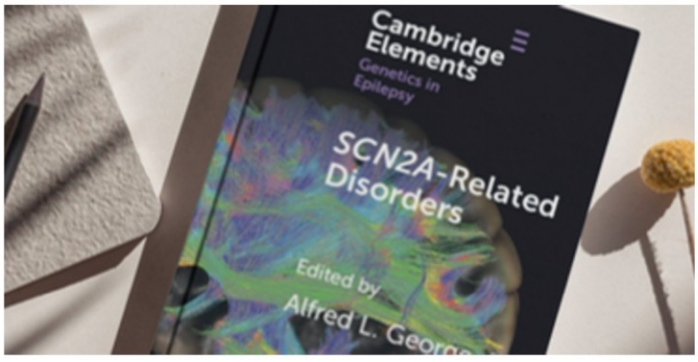
EXPLORE MORE RESOURCES

Individuals with SCN2A-related disorders may benefit from care delivered through a **multidisciplinary** approach, bringing together multiple specialists to support the full range of medical and developmental needs.

Learn more about SCN2A Multidisciplinary Centers →

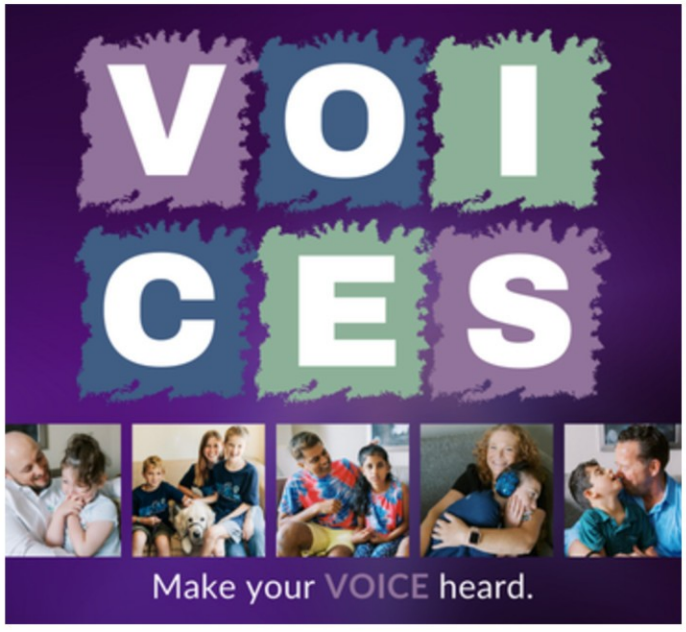
The ***Cambridge Elements Genetics in Epilepsy*** series is a free, peer-reviewed resource written by experts in the field. It provides clear, accessible overviews of SRDs, including genetics, clinical features, and current approaches to care and research. Families and clinicians alike often use this resource to support shared understanding and informed conversations.

The Foundation has made this available online at no cost. →



The **DRAGONFLY Study** is a global natural history study designed to better understand SRDs across the lifespan, helping researchers learn how different SCN2A variants affect development, seizures, and daily life. Participation helps inform future clinical care guidelines and treatment development for the broader SCN2A community.

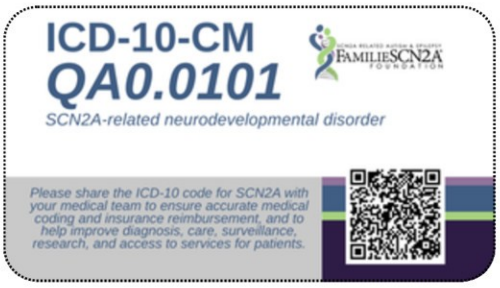
Learn more about the DRAGONFLY Study and register today →



SCN2A now has its own **ICD-10-CM code, QA0.0101**, which supports accurate documentation of SCN2A-related disorders across healthcare systems. Using this specific code helps ensure clearer medical records, supports access to appropriate services and therapies, and improves communication among healthcare providers.

Having a dedicated ICD-10-CM code also strengthens recognition of SCN2A in clinical care, research, and health data, helping inform future care standards and treatment development. Please share this code with your provider at every visit.

Download a copy of your card here



The **FamilieSCN2A Global Support Network** is a private, online community created exclusively for families and caregivers impacted by SRDs. This space allows families from around the world to connect, ask questions, share lived experience, and learn from one another in a supportive environment.

Connect with one of our Parent Navigators and join the network today!

"For the first time, we were surrounded by people who understood our worries, our exhaustion, and our hope—without us having to say a word."

— Sarah, Mom to Calvin.

The **SCN2A Family & Professional Conference** is the world's largest gathering dedicated to SRDs, bringing together families, researchers, clinicians, industry partners, advocates, and volunteers from around the globe.

There is nothing quite like being in a space surrounded by people who truly understand this journey—families sharing lived experience, scientists and clinicians working toward breakthroughs, and a community united by hope and purpose. The conference is a place to learn, connect, and feel renewed, as ideas take root and meaningful relationships are built well beyond the conference walls.
It is a powerful reminder of what is possible when this community comes together.

Learn more here

MEETING FAMILIES WHERE THEY ARE

Our programs are designed to offer connection, practical support, and moments of joy along the way.

At the heart of our work is community. Through our ***Global Support Network***, families from around the world connect in a private, welcoming online space to ask questions, share experiences, and find reassurance from others who truly understand life with SCN2A. Many parents tell us this is where they first felt less alone.

Within that community, our ***Caregivers Connection*** provides a dedicated space for parents and caregivers to share openly, support one another, and focus on self-care with others who understand the demands of caregiving for a child with with an SCN2A-related disorder.

SCN2A affects the whole family, including siblings. ***SuperSibs Club*** is a dedicated space for siblings of individuals with SRDs to connect with peers, enjoy creative activities, and access resources just for them.

Along the way, we also pause to celebrate strength. ***Warrior Wednesday*** honors individuals living with SCN2A by sharing their courage, resilience, and unique journeys—one story at a time.

Because connection matters, both online and close to home, we offer ***Family Event Grants***, small grants that help families host local gatherings, meet-ups, or retreats. These moments build community and remind families they are never alone.

We also offer ***Family Travel Scholarships*** to help offset the costs associated with travel to our Family & Professional Conference, because we believe access to education, connection, and community should not be limited by geography or financial barriers.

Our SCN2A ***Patient Assistance Grants*** help eligible families access essential medical equipment, therapy tools, and educational supports that may not be covered elsewhere, as funding allows.

Together, these programs reflect our commitment to supporting families not only at diagnosis, but throughout their lives.

To explore our programs, visit:
www.scn2a.org/programs-events

YOU'RE NOT ALONE.

Whether you're new to this journey or further along, your questions are valid—and your voice matters.
Understanding your loved one's SCN2A diagnosis and individual presentation can empower you to advocate for your child, connect with others who understand, and make informed decisions.

The FamilieSCN2A Foundation is here to support you
every step of the way.

DID YOU **KNOW?**

SCN2A AFFECTS EACH PERSON DIFFERENTLY

Even people with the same SCN2A variant can have very different strengths, challenges, and support needs.

SCN2A IS A SPECTRUM

Some individuals have epilepsy, some have autism, some have both. Developmental paths vary widely.

SCN2A IS A LIFELONG CONDITION

Most children do not outgrow SCN2A-related disorders, but therapies, supports, and care can change and evolve over time.

EARLY DIAGNOSIS MATTERS

Earlier genetic diagnosis can help guide treatment decisions, support planning, and ongoing monitoring.

TREATMENT AND CARE ARE HIGHLY INDIVIDUALIZED

There is no single treatment plan that works for everyone. Care decisions are best made in partnership with your loved one's medical team.

FAMILIES ARE NOT ALONE—AND THEIR VOICES MATTER.

Families play a vital role in research, care, and advocacy, and a global SCN2A community is here to support you.





QUESTIONS TO ASK YOUR CARE TEAM

UNDERSTANDING THE GENETIC VARIANT

- What is my loved one's specific genetic variant?
- What type of variant is it (for example, missense, truncation, deletion, or duplication)?
- Is this variant considered pathogenic (disease-causing)?
- What is the functional effect of this variant (gain-of-function, mixed-function or loss-of-function)?
  - Is this based on functional studies or prediction tools?
- Does my loved one have any additional genetic variants?
- What is the inheritance pattern of this variant, and what does that mean for our family?
- Is additional genetic testing recommended (such as parental testing or exome sequencing)?

UNDERSTANDING THIS CONDITION

- Based on this specific variant, what clinical features might we expect to see in our loved one?
- How might this variant affect development, seizures, behavior, or learning?

CARE, TREATMENT, AND PLANNING

- What medications or therapies are appropriate for my loved one?
- Are there any medications that should be avoided?
- With an SRD diagnosis and seizure history, what are the risks of life-threatening outcomes, and how can we reduce it?
- How do we create a Seizure Action Plan?
- What other specialists should be involved in my loved one's care?

RESEARCH, EXPERTISE, AND CONNECTION

- Are there clinical trials or research studies my loved one may be eligible for?
- Has my loved one's variant been reported before?
- Do you feel comfortable managing this condition, or would you be willing to collaborate with an SCN2A specialist?

COMMENTS/ NOTES

Learn more about the FamilieSCN2A Foundation!
Visit **SCN2A.ORG**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