

# 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

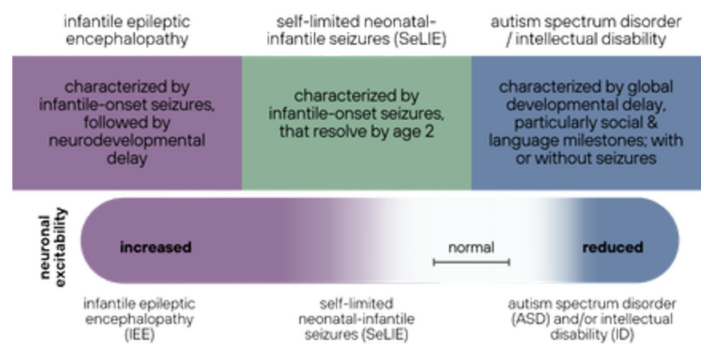
Changes in SCN2A are among the most common causes of neurodevelopmental disorders. Even in people with the same variants, clinical presentation may vary.



## 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., Abbot, 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>