SCN2A-RELATED DISORDERS (SRD)

QUICK FACT SHEET

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 nerve impulses are conducted.

SCN2A

SODIUM CHANNEL NUMBER 2 ALPHA SUBNIT

ASSOCIATED MEDICAL CONDITIONS

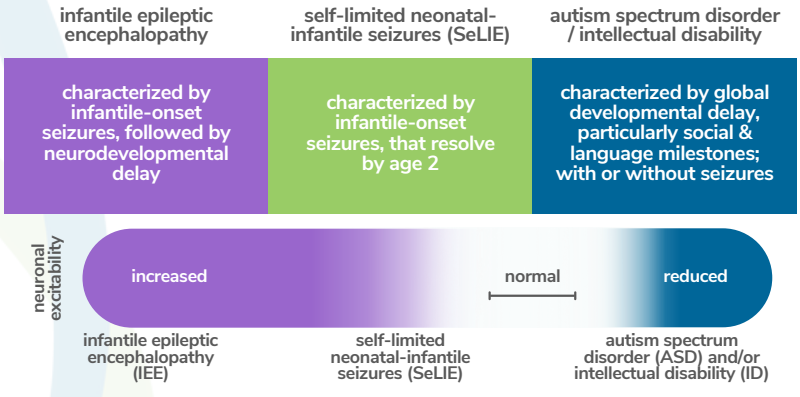
Epilepsy, Autism Spectrum Disorder, Attention Deficit Hyperactivity Disorder, Global Developmental Delays, Intellectual Disability, Movement & Speech Disorders, Gastrointestinal & Urology Issues, Cortical Visional Impairment, Sleep Issues, Dysautonomia, Dystonia, Feeding Issues, Neuropathic Pain, Ataxia and Cerebral Palsy

Patients with SCN2A are seen by a variety of specialists to address their specific medical challenges and benefit from a multi-disciplinary team approach to deliver comprehensive care.

SCN2A is one of the most common causes of neurodevelopmental disease. Even in variants that are repeated within SCN2A, presentation may vary.

How does SCN2A Present?

The current published literature suggests two main presentations. A gain of function variant makes the channel more excitable, typically leading to infantile-onset seizures. While a loss of function variant reduces the excitability or destroys channel function altogether, typically leading to autism spectrum disorder and/or intellectual disability.



SCN2A-RELATED AUTISM & EPILEPSY
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