

SCN2A-RELATED AUTISM

HOW IS SCN2A RELATED AUTISM DIFFERENT?

SCN2A related autism can present both with and without epilepsy. Currently there is not a specific phenotype (or presentation) seen with SCN2A related autism. Individuals with Autism Spectrum Disorder (ASD) tend to have challenges in these three specific areas:

- Social Interactions
- Communication
- Restricted, Repetitive & Stereotyped Patterns of Behavior

Additionally, in SCN2A related autism, challenges with motor planning and gastrointestinal issues have been reported.

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?

It is estimated that there will be approximately 11 SCN2A-related cases per 100,000 births.

Over 400 SCN2A-mediated disorders children will be born each year in the United States alone.

Incidence of loss-of-function cases are expected to be approximately five-fold higher than gain-of-function cases.

Source: Sanders, et al. (2018). Progress in understanding and treating SCN2A-mediated disorders. *Trends in Neuroscience*, 41(7):442-456.

QUICK FACT SHEET

LOSS OF FUNCTION IN SCN2A

In contrast to gain of function variants that contribute to seizure, 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.

Ben-Shalom, et al. Opposing effects on NaV1.2 function underlie differences between SCN2A variants observed in individuals with autism spectrum disorder or infantile seizures. *Biological Psychiatry*, 2017



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.