

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.