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.

**HOW RARE IS SCN2A?**

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.

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.


**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*

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