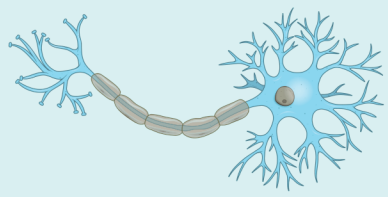
WHAT ARE POSSIBLE CAUSES OF AUTISM AND EPILEPTIC SEIZURES AT A GENETIC LEVEL?

Important Terms:

gene: a building block of the body that are instruction manuals or a recipe book; errors in the gene sequence are called **mutations**

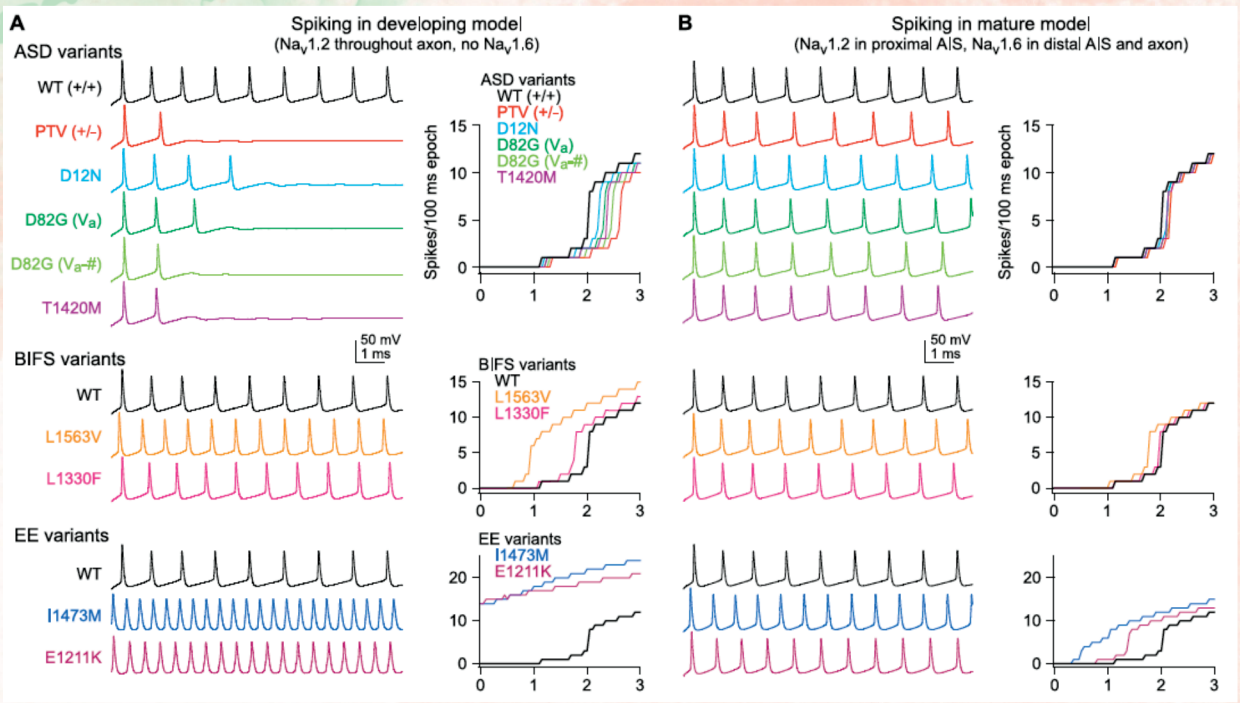


neuron: electrically excitable cells that are responsible for transmitting signals in the brain

For the last 50 years, SCN2A gene was known to be linked to epilepsy while more recent studies show it is also linked to autism.

SCN2A gene translates to a neuronal sodium channel which is an essential electrical component. Mutations in this gene disrupt neuronal activity.

In this study, researchers simulated in a computer how mutations in the SCN2A gene disrupts neuronal electrical activity, which could ultimately cause autism and epilepsy.



autism (ASD)	*epilepsy (EE)*
less firing of neurons	*more firing of neurons*
less potential to make connections	*more potential for unnecessary connections*

Mutations in the SCN2A gene may lead to autism or epileptic seizures.

Ben-Shalom, R., Keeshen, C. M., Berrios, K. N., An, J. Y., Sanders, S. J., & Bender, K. J. (2017). Opposing Effects on NaV1.2 Function Underlie Differences Between SCN2A Variants Observed in Individuals With Autism Spectrum Disorder or Infantile Seizures. Biological psychiatry, 82(3), 224–232. https://doi.org/10.1016/j.biopsych.2017.01.009