# Molecular ‘Rosetta Stone’ could provide insight into autism

Distinct sets of genetic defects in a single neuronal protein can lead either to infantile epilepsy or to autism spectrum disorders (ASDs), depending on whether the respective mutations boost the protein’s function or sabotage it, according to a new study by UC San Francisco researchers.

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In studies published in 2012, 2014, and 2015, [researchers] found that *de novo* genetic mutations — spontaneous mutations not inherited from parents — play a role in the development of ASDs in at least 10 percent of all cases of autism, many more than previously recognized.

These studies led to the identification of 65 genes with a strong likelihood of contributing to autism when mutated and confirmed SCN2A as one of the top hits.

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This study represents a first step in understanding how SCN2A mutations lead to autism and developmental delay….

“These findings solidify *SCN2A*’s status as one of the most important genes in autism,” said [neurophysiologist Kevin Bender, an assistant professor of neurology]. “They give us a place to start exploring exactly how changes in early brain development lead to this condition.”

[The study can be found here.]

**The GLP aggregated and excerpted this blog/article to reflect the diversity of news, opinion, and analysis. Read full, original post:** Autism Researchers Discover Genetic ‘Rosetta Stone’