

# Single mutation causes seizure disorder

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Lennox-Gastaut syndrome is a severe early-onset seizure disorder that includes cognitive and behavioral abnormalities. Its pathophysiology is largely unknown.

Recent whole-exome sequencing in patients with Lennox-Gastaut syndrome identified a [spontaneous mutation](#) in the gene encoding a GABA-A receptor subunit implicated in other childhood epilepsies.

Robert Macdonald, MD, PhD, and colleagues have now generated a mouse model with the human mutation in the GABA-A receptor subunit gene. The mice have spontaneous atypical absence seizures and other types of seizures, with EEG characteristics consistent with Lennox-Gastaut syndrome. The mice also have abnormal behaviors observed in patients with Lennox-Gastaut syndrome, including impaired learning, memory, and [social interaction](#), increased anxiety and hyperactivity.

The findings, reported in the journal *Brain Communications*, demonstrate that a single mutation in one gene can impair inhibitory GABA signaling in the brain and cause multiple types of seizures and behavioral abnormalities. The [mouse model](#) will be useful for further characterization of Lennox-Gastaut syndrome pathophysiology and for the development of novel therapies.

**More information:** Shimian Qu et al. GABAA receptor  $\beta 3$  subunit mutation D120N causes Lennox–Gastaut syndrome in knock-in mice, *Brain Communications* (2020). [DOI: 10.1093/braincomms/fcaa028](https://doi.org/10.1093/braincomms/fcaa028)

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