

# Researchers identify genetic cause of MMPSI, rare infant epileptic disorder

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Credit: AI-generated image ([disclaimer](#))

A Yale-led team of researchers has identified the gene that, when mutated, causes a devastating early-onset disorder in babies known as "malignant migrating partial seizures of infants," or MMPSI. The study appears online in *Nature Genetics*.

MMPSI is a rare, severe brain disorder that appears in the first six months of life. It is characterized by treatment-resistant epileptic seizures and developmental delay. Further, the electrical activity appears to "migrate" through various regions of the brain, which gives the disorder its name. Although the [recurrent seizures](#) and electrical discharges in the brain abate with age, the patient's intellectual and motor development is arrested in infancy.

In collaboration with a group at the Hopital Necker-Enfants Malades in Paris, France, the Yale team found that the disease-causing gene, which was identified by selective DNA exome sequencing in a dozen patients affected by MMPSI, results in the overactivity of a protein that normally controls the excitability of neurons. The Yale team also discovered that the affected protein interacts with another protein that, when defective, is known to produce [Fragile X syndrome](#), the leading inherited cause of autism and intellectual disability.

"For the first time, we have a target for future therapeutic approaches to treating this devastating condition," said co-first author Matthew Fleming, [postdoctoral researcher](#) at Yale School of Medicine.

All of the data to characterize the aberrant protein in this study were generated in lab of Leonard Kaczmarek, professor of pharmacology and of cellular and molecular physiology at Yale School of Medicine.

Kaczmarek explains, "We believe this to be the first epilepsy-associated gene to be identified by exome sequencing in a handful of affected patients," says Kaczmarek. "Moreover, it provides a very important glimpse into why some epilepsies are so devastating for brain function while others seem to have no effect on intellectual development."

**More information:** [www.nature.com/ng/journal/vaop...nt/full/ng.2441.html](http://www.nature.com/ng/journal/vaop...nt/full/ng.2441.html)

Provided by Yale University

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