■ Case vignette

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Epileptic Disorders

The epilepsy-movement disorder phenotypic spectrum and phenytoin-induced dyskinesia associated with *GABRB3* pathogenic variants

Amaar Marefi[1], Myriam Srour[1,2]

[1] Division of Pediatric Neurology, Departments of Pediatrics and Neurology & Neurosurgery, McGill University, Montreal, Quebec, Canada

[2] McGill University Health Center Research Institute, Montreal, Quebec, Canada

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- This manuscript sheds light on the broad spectrum of epilepsy-movement disorder phenotypes and provides evidence that genes encoding GABA A receptor subunits, such as *GABRB3, GABRA2, GABRG2*, and *GABRB2* are implicated in pathogenesis.

- 10 previously reported patients found to have pathogenic variants in the *GABRB3* gene had a movement disorder in addition to epilepsy.

- These patients presented with various movement disorders including choreoathetosis, dystonia, dyskinesia, hand stereotypies, tremors, myoclonus, and hypotonia.

- Children with epileptic encephalopathy and patients with pre-existing dyskinesias are more susceptible to develop phenytoin-induced dyskinesia.

- This suggests that genetic factors, such as *GABRB3* pathogenic variants, may represent a susceptibility to phenytoin-induced dyskinesia.

- We provide a video of a patient, known to have a *de novo* pathogenic *GABRB3* variant, who developed dyskinesias of the mouth, head and hands soon after receiving a phenytoin load.

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