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## ***NEXMIF* Epilepsy: An Alternative Cause of Progressive Myoclonus**

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**Contributions:**

Lauren E Chorny: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Additional contributions: Literature review - Literature Review - Douglas R Nordli III

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## Case Summary

An 8 year old boy with generalized myoclonic epilepsy followed by progressive cognitive decline presented with worsening myoclonus despite being compliant with prescribed clobazam. The movements (Video 1) in conjunction with a worsening cognitive status over time were concerning for a progressive myoclonic epilepsy. Initial EEG captured frequent myoclonic seizures time-locked with spike-wave activity (Figure). Overnight EEG revealed normal sleep architecture. His seizures stopped with valproic acid load. Genetic testing revealed a heterozygous pathogenic variant in NEXMIF (c.2478\_2479dup), which is associated with NEXMIF encephalopathy. NEXMIF encephalopathy is characterized by mild to severe intellectual disability and includes myoclonic seizures, absence seizures and atonic seizures.<sup>1</sup>

Traditionally, the differential diagnosis of progressive myoclonic epilepsy entails diseases such as Lafora body disease, Unverricht-Lundborg disease, NCL, Type 1 Sialidosis and MERRF.<sup>2</sup> This case emphasizes the consideration of NEXMIF mutations in the differential diagnosis of a suspected progressive myoclonic epilepsy.

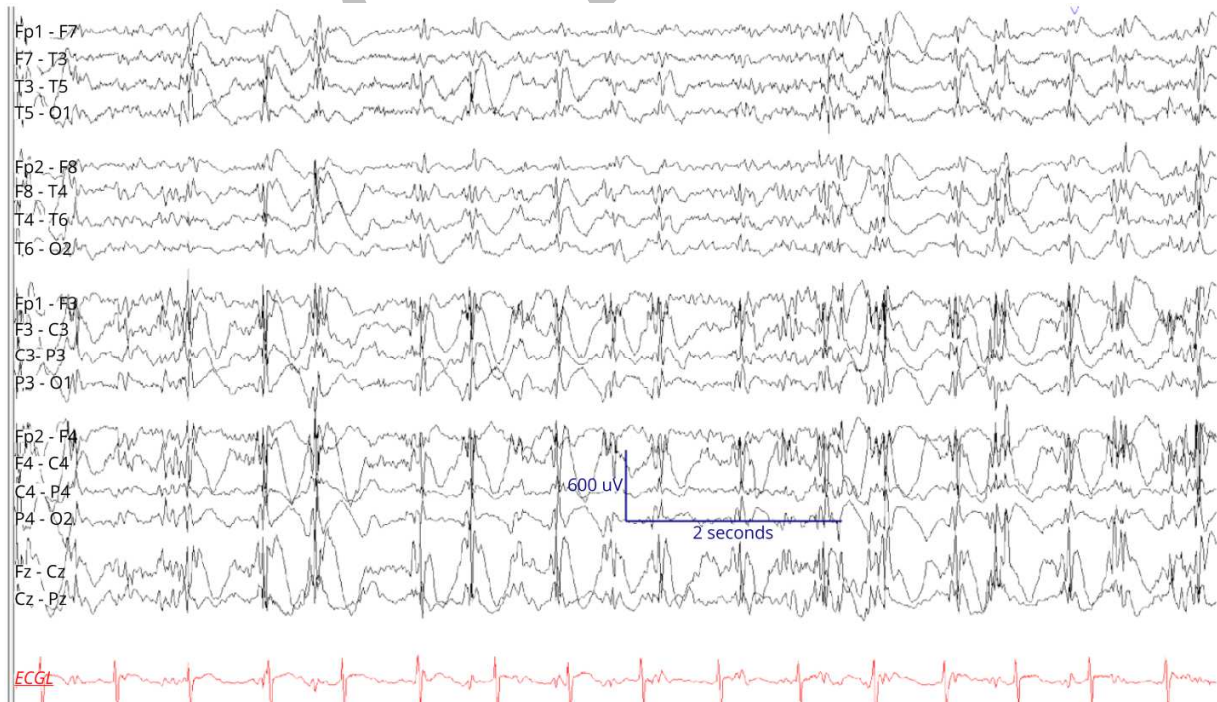
## Figure Legends

### Video 1:

Video of myoclonus.

### Figure:

Longitudinal bipolar montage EEG with diffuse spike wave associated with myoclonus.



<http://links.lww.com/WNL/C544>

**References:**

1. Stamberger, Hannah et al. "NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns." *Genetics in medicine : official journal of the American College of Medical Genetics* vol. 23,2 (2021): 363-373. doi:10.1038/s41436-020-00988-9
2. Girard, Jean-Marie, et al. "Progressive myoclonus epilepsy." *Handbook of clinical neurology* 113 (2013): 1731-17

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