NEXMIF Epilepsy: An Alternative Cause of Progressive Myoclonus

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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.  

Traditionally, the differential diagnosis of progressive myoclonic epilepsy entails diseases such as Lafora body disease, Unverricht-Lundbord disease, NCL, Type 1 Sialidosis and MERRF.  

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.
References:
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