
**Format**: Abstract


**Epilepsy in LAMA2-related muscular dystrophy: An electro-clinico-radiological characterization.**


**Author information**

**Abstract**

**OBJECTIVE**: To delineate the epileptic phenotype of LAMA2-related muscular dystrophy (MD) and correlate it with the neuroradiological and muscle biopsy findings, as well as the functional motor phenotype.

**METHODS**: Clinical, electrophysiological, neuroradiological, and histopathological data of 25 patients with diagnosis of LAMA2-related MD were analyzed.

**RESULTS**: Epilepsy occurred in 36% of patients with LAMA2-related MD. Mean age at first seizure was 8 years. The most common presenting seizure type was focal-onset seizures with or without impaired awareness. Visual aura and autonomic signs, including vomiting, were frequently reported. Despite a certain degree of variability, bilateral occipital or temporo-occipital epileptiform abnormalities were by far the most commonly observed. Refractory epilepsy was found in 75% of these patients. Epilepsy in LAMA2-related MD was significantly more prevalent in those patients in whom the cortical malformations were more extensive. In contrast, the occurrence of epilepsy was not found to be associated with the patients' motor ability, the size of their white matter.
abnormalities, or the amount of residual merosin expressed on muscle.

**SIGNIFICANCE:** The epileptic phenotype of LAMA2-related MD is characterized by focal seizures with prominent visual and autonomic features associated with EEG abnormalities that predominate in the posterior quadrants. A consistent correlation between epileptic phenotype and neuroimaging was identified, suggesting that the extension of the polymicrogyria may serve as a predictor of epilepsy occurrence.

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**KEYWORDS:** LAMA ; EEG; MDC1A; epilepsy; merosin; seizures

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