Presentation of the Second Case in the World of “Cortıcal Myoclonıc Tremor with epilepsy, Familial 7”

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Introduction:

Familial Adult Myoclonic Epilepsy (FAME) is an infrequent autosomal dominant disorder characterized by the onset of myoclonus, generalized tonic-clonic seizures, and cortical tremor during adolescence or adulthood.

RAPGEF-2 mutation-related myoclonic epilepsy –which defined as Cortıcal Myoclonıc Tremor Wıth Epılepsy, Famılıal, 7 is reported in 1 patient in the World.

Case:

A 17-year-old girl had been given a preliminary diagnosis of laphora or late onset NCL. She presented with myoclonus at the age of 10 and has since developed a slowly progressive myoclonic movement disorder triggered by motion. There is no family history of the condition. She had high lumbar puncture pressure in history, for this she used acetazolamide. On examination, she exhibited variable levels of ataxia and myoclonus. Cardiac, abdominal, and endocrine evaluations were normal. The patient's psychometric evaluation yielded exceptional results. Whole exome sequencing was performed at two different centers for juvenile myoclonic epilepsy, but the results were inconclusive. The patient experiences movement-triggered myoclonus with varying frequency, ranging from once a week to multiple times a day, resulting in drop attacks. The patient's myoclonus during examination hindered her ability to walk. However, subsequent EEGs revealed a slow spike-wave associated with myoclonus. The patient was diagnosed with RAPGEF-2 mutation-related myoclonic epilepsy, Familial, 7 which is only the second case that will be reported. Despite treatment with valproate and zonisamide being ineffective, had partial benefit from clobazam therapy.

Conclusion:

Cortical tremors pose a challenge in distinguishing myoclonus.(1,2) In the presence of tremulous, stress-induced myoclonic seizures beginning in adolescence or later and medically refractory seizures, autosomal dominant FAME should be considered.(1,3) FAME has a slow, progressive clinical course occurring with intellectual disability and worsening of both tremor and myoclonus although with a less severe decline compared to other progressive myoclonic epilepsies. Valproate, levetiracetam, and benzodiazepines are considered the first-line treatments. Advanced genetic tests should be performed and detailed information about these patients results should be published to obtain more

References

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