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Oral Presentation

Juvenile Myoclonic Epilepsy as A Spectrum Disorder

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Abstract

In consequence of newer research juvenile myoclonic epilepsy (JME) is no longer seen as a homogeneous disease. The causes of the existing variance are only partially known yet. We discuss to what extent the phenotypical spectrum of this polygenetically determined disorder expresses genetically defined endophenotypes, or is due to mere quantitative differences in the expression of the core phenotype. Of the three common seizure types of JME, myoclonic, generalized tonic-clonic and absences, absences also occur independently and are strong candidates for an endophenotype. Focal features may in some patients be seen in clinical seizures or the EEG but rarely in both. They have no morphological correlates. In a system epilepsy, local manifestations are possible, and some are due to reflex mechanisms. Of the four reflex epileptic traits common in JME, photosensitivity and praxis induction appear related to basic mechanisms of the core syndrome, whereas language-induced orofacial reflex myocloni and eye closure sensitivity are also seen in other clinical contexts and therefore seem to represent endophenotypes. Cognitive abnormalities indicating slight frontal lobe dysfunction seem to be ubiquitous in JME and are also seen in unaffected siblings of patients. Cluster B personality disorder is found in 1/3 of patients, representing a more severe expression of the underlying pathology. Treatment response and prognosis seem to be affected by an interplay of the described factors producing the severest end of the JME spectrum. The spectrum appears to be due to an interaction of stronger or weaker expression of the core phenotype with various endophenotypes.

Keywords: Epilepsy, Treatment, System.

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