Pediatric Neurology Part I: Chapter 65. Dravet syndrome (severe myoclonic epilepsy in infancy) (Handbook of Clinical Neurology)

Charlotte Dravet, Hirokazu Oguni

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Severe myoclonic epilepsy in infancy (SMEI) is a rare disease, characterized by febrile and afebrile, generalized and unilateral, clonic or tonic–clonic seizures that occur in the first year of life in an otherwise apparently normal infant. They are later associated with myoclonus, atypical absences, and partial seizures. Developmental delay becomes apparent within the second year of life and is followed by definite cognitive impairment and personality disorders of variable intensity. In the borderline form, children do not present with myoclonic symptoms but have the same general picture. SMEI is a channelopathy and the genetic studies have shown a mutation in the SCN1A gene in 70 to 80% of the patients, including the borderline forms. At present, there are no well-established correlations between genotype and phenotype. The electroencephalograms, often normal at the onset, display both generalized and focal anomalies, without a specific electroencephalographic pattern. As a rule, neuroimaging is normal. All seizure types are resistant to antiepileptic drugs and status epilepticus is frequent. Some drugs have been shown to aggravate the seizures and must be avoided. Two recent drugs have been proved to partially control the convulsive seizures and the status epilepticus. Therefore, it is crucial to diagnose this epilepsy soon after its onset in order to prescribe the most appropriate treatment.

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