Pediatric Neurology Part III: Chapter 151. Congenital myasthenic syndromes (Handbook of Clinical Neurology)

Bruno Eymard, Daniel Hantaï, Brigitte Estournet

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Congenital myasthenic syndromes (CMS) are a heterogeneous group of disorders caused by genetic defects affecting neuromuscular transmission and leading to muscle weakness accentuated by exertion. The characterization of CMS comprises two complementary steps: establishing the diagnosis and identifying the pathophysiological type of CMS. The combination of clinical, electrophysiological, and morphological studies allows the physician to refer a given CMS to mutation(s) in one of the 18 causative genes discovered to date and, in turn, to classify the CMS according to the location of the mutated proteins at the neuromuscular junction into presynaptic compartment, synaptic basal lamina, and postsynaptic compartment CMS. This complete characterization is essential for counseling and therapy of the patient, depending on the molecular background of the respective CMS. Despite comprehensive characterization, the phenotypic expression of one given gene involved is variable, and the etiology of many CMS remains to be discovered.

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