The unfolding landscape of the congenital myasthenic syndromes.

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Abstract

Congenital myasthenic syndromes (CMS) are heterogeneous disorders in which the safety margin of neuromuscular transmission is impaired by one or more specific mechanisms. Since the advent of next-generation sequencing methods, the discovery of novel CMS targets and phenotypes has proceeded at an accelerated rate. Here, we review the current classification of CMS and describe our findings in five of these targets identified and investigated in our laboratory in the past 5 years. Defects in LRP4 hinder synaptic development and maintenance; the defects in PREPL are predicted to diminish filling of the synaptic vesicle with acetylcholine; and defects in SNAP25, Munc13-1, and synaptotobrevin-1 impede synaptic vesicle exocytosis.

KEYWORDS: LRP4; Munc13-1; PREPL; SNAP25B; congenital myasthenic syndromes; synaptobrevin


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