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Congenital myasthenic syndromes with acetylcholinesterase deficiency, the pathophysiological mechanisms.

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Abstract

The neuromuscular junction (NMJ) is a cholinergic synapse in vertebrates. This synapse connects motoneurons to muscles and is responsible for muscle contraction, a physiological process that is essential for survival. A key factor for the normal functioning of this synapse is the regulation of acetylcholine (ACh) levels in the synaptic cleft. This is ensured by acetylcholinesterase (AChE), which degrades ACh. A number of mutations in synaptic genes expressed in motoneurons or muscle cells have been identified and are causative for a class of neuromuscular diseases called congenital myasthenic syndromes (CMSs). One of these CMSs is due to deficiency in AChE, which is absent or diffuse in the synaptic cleft. Here, I focus on the origins of the syndrome. The role of ColQ, a collagen that anchors AChE in the synaptic cleft, is discussed in this context. Studies performed on patient biopsies, transgenic mice, and muscle cultures have provided a more comprehensive view of the connectome at the NMJ that should be useful for understanding the differences in the symptoms observed in specific CMSs due to mutated proteins in the synaptic cleft.

KEYWORDS: ColQ; MuSK; acetylcholine receptor; acetylcholinesterase; congenital myasthenic syndromes; neuromuscular junction

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