PHENOTYPIC CLINICAL CLUES TO DIAGNOSIS OF CONGENITAL MYASTHENIC SYNDROMES						
Phenotype Clue	**AChE deficient**	**ChAT deficient**	**AChR deficient**	**Dok7 myasthenia**	**Rapsyn myasthenia**	**Glycosylation deficits**
Dominant inheritance			SC			
Selective weakness dorsal forearm			SC			
Predominantly limb-girdle distribution				X		X
Congenital contractures		X	X		X	
Sudden apneic episodes provoked by fever or stress		X			X	
Worsened or refractory with AChE inhibitors	X			X		
Repetitive CMAP	X		SC			
Induced by subtetanic stimulation followed by slow recovery		X				

Abbreviations: AChE, acetylcholinesterase; AChR, acetylcholine receptor; ChAT, choline acetyltransferase; CMAP, change in muscle action potential; SC slow channel. syndrome.

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