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## Sarcomeric myopathies associated with tremor: new insights and perspectives

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### Abstract

Myopathies are a large and heterogeneous group of disorders associated with mutations in structural and regulatory genes responsible for proper muscle assembly, organization and function. Despite the molecular diversity of inherited myopathies, they have historically been classified by the phenotypic traits observed in affected patients. It is therefore common for myopathies originating from mutations in different genes to be grouped together due to similar physical manifestations, and conversely myopathies resulting from mutations in the same gene to be considered separately due to disparate symptoms. Herein, we focus on an early onset myopathy linked to inherited or de novo mutations in sarcomeric genes that is characterized by muscle weakness, hypotonia and tremor, and further highlight that it may constitute a new form of myopathy, with tremor as its defining feature. Based on recent reports, we also discuss the possible myogenic origin of the tremor that may start at the level of the sarcomere due to structural and/or contractile alterations occurring as a result of the identified mutations. It is our hope that establishment of this form of myopathy accompanied by myogenic tremor as a new disease entity will have important diagnostic and therapeutic implications.

**Keywords:** Congenital myopathy; Hypotonia; Muscle weakness; Sarcomeric genes; Tremor.

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