Untangling the Complexity of Limb-girdle Muscular Dystrophies.

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Abstract

Limb-girdle muscular dystrophies (LGMD) are a group of genetically heterogeneous, autosomal inherited muscular dystrophies with childhood-to-adult onset, manifesting with hip and shoulder girdle muscle weakness. When the term LGMD was first conceptualized in 1954, it was thought to be a single entity. Currently, there are 8 autosomal dominant (LGMD1A-1H) and 26 autosomal recessive (LGMD2A-2Z) variants according to the Online Mendelian Inheritance in Man database. In addition, there are other genetically-identified muscular dystrophies with a LGMD phenotype not yet classified as LGMD. This highlights the entanglement of LGMD, which represents an area in continuous expansion. Here we aim to simplify the complexity of LGMD by subgrouping them on the basis of the underlying defective protein and its impaired function. This article is protected by copyright. All rights reserved.

KEYWORDS: LGMD; calpainopathy; caveolae-associated muscular dystrophies; limb-girdle muscular dystrophies; muscular dystrophies with defective membrane repair; myofibrillar myopathy; nuclear envelopathies; sarcoglycanopathies; z-disk proteinopathies; α-dystroglycanopathies

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