The Italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis.


Abstract

INTRODUCTION: Limb girdle muscular dystrophies (LGMDs) are characterized by high molecular heterogeneity, clinical overlap, and a paucity of specific biomarkers. Their molecular definition is fundamental for prognostic and therapeutic purposes.

METHODS: We created an Italian LGMD registry that included 370 molecularly defined patients. We reviewed detailed retrospective and prospective data and compared each LGMD subtype for differential diagnosis purposes.

RESULTS: LGMD types 2A and 2B are the most frequent forms in Italy. The ages at disease onset, clinical progression, and cardiac and respiratory involvement can vary greatly between each LGMD subtype. In a set of extensively studied patients, targeted next-generation sequencing (NGS) identified mutations in 36.5% of cases.

CONCLUSION: Detailed clinical characterization combined with muscle tissue analysis is fundamental to guide differential diagnosis and to address molecular tests. NGS is useful for diagnosing forms without specific biomarkers, although, at least in our study cohort, several LGMD disease
although, at least in our study cohort, several LGMD disease mechanisms remain to be identified. Muscle Nerve 55: 55-68, 2017.

© 2016 Wiley Periodicals, Inc.

PMID: 27184587 [Indexed for MEDLINE]

Full text

Full text at journal site