Limb-girdle muscular dystrophies - international collaborations for translational research.

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Abstract

The limb-girdle muscular dystrophies (LGMDs) are a diverse group of genetic neuromuscular conditions that usually manifest in the proximal muscles of the hip and shoulder girdles. Since the identification of the first gene associated with the phenotype in 1994, an extensive body of research has identified the genetic defects responsible for over 30 LGMD subtypes, revealed an increasingly varied phenotypic spectrum, and exposed the need to move towards a systems-based understanding of the molecular pathways affected. New sequencing technologies, including whole-exome and whole-genome sequencing, are continuing to expand the range of genes and phenotypes associated with the LGMDs, and new computational approaches are helping clinicians to adapt to this new genomic medicine paradigm. However, 60 years on from the first description of LGMD, no curative therapies exist, and systematic exploration of the natural history is still lacking. To enable rapid translation of basic research to the clinic, well-phenotyped and genetically characterized patient cohorts are a necessity, and appropriate outcome measures and biomarkers must be developed through natural history studies. Here, we review the international collaborations that are addressing these translational research issues, and the lessons learned from large-scale LGMD sequencing programmes.

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