Muscular dystrophies.

Mercuri E¹, Bönnemann CG², Muntoni F³.

Author information

1 Pediatric Neurology Unit, Università Cattolica del Sacro Cuore Roma, Rome, Italy; Nemo Clinical Centre, Fondazione Policlinico Universitario A Gemelli IRCCS, Rome, Italy.

2 Neuromuscular and Neurogenetic Disorders of Childhood Section, National Institute of Neurological Disorders and Stroke, National Institutes of Health, Bethesda, MD, USA.

3 Dubowitz Neuromuscular Centre, University College London, Great Ormond Street Institute of Child Health, London, UK; National Institute for Health Research Great Ormond Street Hospital Biomedical Research Centre, London, UK. Electronic address: f.muntoni@ucl.ac.uk.

Abstract

Muscular dystrophies are primary diseases of muscle due to mutations in more than 40 genes, which result in dystrophic changes on muscle biopsy. Now that most of the genes responsible for these conditions have been identified, it is possible to accurately diagnose them and implement subtype-specific anticipatory care, as complications such as cardiac and respiratory muscle involvement vary greatly. This development and advances in the field of supportive medicine have changed the standard of care, with an overall improvement in the clinical course, survival, and quality of life of affected individuals. The improved understanding of the pathogenesis of these diseases is being used for the development of novel therapies. In the most common form, Duchenne muscular dystrophy, a few personalised therapies have recently achieved conditional approval and many more are at advanced stages of clinical development. In this Seminar, we concentrate on clinical manifestations, molecular pathogenesis, diagnostic strategy, and therapeutic developments for this group of conditions.

Copyright © 2019 Elsevier Ltd. All rights reserved.

PMID: 31789220 DOI: 10.1016/S0140-6736(19)32910-1