
Format: Abstract


LMNA-Related Muscular Dystrophy with Clinical Intrafamilial Variability.

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Abstract
The LMNA gene is associated to a huge broad of phenotypes, including congenital Emery-Dreifuss muscular dystrophy and late-onset LMNA-related muscular dystrophy. In these forms, muscle weakness, contractures, and cardiac impairment are common. In an autosomal dominant pedigree including 5 affected patients, NGS molecular analysis performed in 6 relatives identifies the heterozygous c.1129C>T p.Arg377Cys variant in the exon 6 of the LMNA gene in three of them. Clinical, laboratorial, imaging investigation of these affected patients showed a significant clinical variability: the father presented subclinical imaging muscular dystrophy masqueraded as radiculopathy. One of his sons presented cardiac arrhythmia, muscular weakness, elbow contractures, and intranuclear pseudoinclusions on muscle biopsy. A second son presented only decreased tendon reflexes. Two other brothers presenting myalgia and cramps were not carriers of the same mutation in the LMNA gene. Early diagnosis, considering these variable phenotype and genotype, is important for genetic counseling, as well as cardiac, and rehabilitation management.

KEYWORDS: Electron microscopy; Imaging; LMNA; Muscle biopsy; Muscular dystrophy; Radiculopathy

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