Muscle fiber type disproportion (FTD) in a family with mutations in the LMNA gene.


Abstract

INTRODUCTION: Mutations in the lamin A/C protein cause laminopathies, a heterogeneous group of disorders that include recessive axonal neuropathy (CMT2B1), Emery-Dreifuss muscular dystrophy (EDMD), limb-girdle muscular dystrophy (LGMD), dilated cardiomyopathy with conduction defect, and different forms of lipodystrophy and progeria.

METHODS: We provide clinical, histopathological, muscle imaging, and cardiac features of a family with heterozygous mutation in the LMNA gene.

RESULTS: We identified heterozygous mutations (c.80C> T; pT27I) in the LMNA gene in 3 family members who had the LGMD phenotype with onset in their early thirties and cardiac conduction defects or dilated cardiomyopathy. Interestingly, muscle biopsies showed changes consistent with fiber type disproportion (FTD).

CONCLUSIONS: Fiber type disproportion has been reported only anecdotally in muscle biopsies of patients with LMNA mutations. Our report further supports this association and suggests inclusion of molecular testing for LMNA in the differential diagnosis of myopathies with FTD due to the risk for life threatening events.

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KEYWORDS: FTD; LGMD; LMNA; cardiomyopathy; laminopathy

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