Emery-Dreifuss muscular dystrophy type 2 associated (?) with mild peripheral polyneuropathy.


Abstract

In recent years numerous mutations in the LMNA gene encoding lamin A/C were shown to segregate with a wide spectrum of phenotypes. A recurrent p.R377H mutation in the LMNA gene was reported in patients with Emery-Dreifuss dystrophy (EDMD2) with various ethnic backgrounds. We present a patient with EDMD2 caused by a p.R377H mutation, associated with mild peripheral polyneuropathy. The analysis of peripheral myelin protein 22 (PMP22), ganglioside induced differentiation-associated protein 1 (GDAP1), gap junction β-1 protein (GJB1), and myelin protein zero (MPZ) genes did not reveal mutations; however, we identified a new sequence intronic variant in the mitofusin 2 (MFN2) gene of unknown pathogenic significance. A complex phenotype in the presented patient might depend either on single mutation in the LMNA gene or on bigenic defect; therefore, a wide genetic investigation is needed to elucidate the molecular background of EDMD2/polyneuropathy in this case.

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