Unclassifiable arrhythmic cardiomyopathy associated with Emery-Dreifuss caused by a mutation in FHL1.


Abstract
Emery-Dreifuss muscular dystrophy (EDMD) is a heterogeneous genetic disorder characterized by peripheral muscular weakness often associated with dilated cardiomyopathy. We characterize clinically a large family with a mutation in FHL1 gene (p.Cys255Ser). Penetration was 44%, 100% for males and 18% for females. The heart was the main organ involved. Affected adult males had mild hypertrophy, systolic dysfunction and restriction with non-dilated ventricles. Carriers had significant QTc prolongation. The proband presented with resuscitated cardiac arrest. There were two transplants. Pathological study of explanted heart showed fibrofatty replacement and scarring consistent with arrhythmogenic cardiomyopathy and prominent left ventricular trabeculations. Myopathic involvement was evident in all males. Females had no significant neuromuscular disease. Mutations in FHL1 cause unclassifiable cardiomyopathy with coexisting EDMD. Prognosis is poor and systolic impairment and arrhythmias are frequent. Thrombopenia and raised creatine phosphokinase should raise suspicion of an FHL-1 disorder in X-linked cardiomyopathy.

KEYWORDS: Emery-Dreifuss muscular dystrophy; FHL1; cardiomyopathy; mutation

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