Limb-girdle muscular dystrophy with severe heart failure overlapping with lipodystrophy in a patient with LMNA mutation p.Ser334del.


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Abstract
Laminopathies, a group of heterogeneous disorders associated with lamin A/C gene (LMNA) mutations, encompass a wide spectrum of clinical phenotypes, which may present as separate disease or as overlapping syndromes. We describe a 35-year-old female in whom a novel sporadic heterozygous mutation c.1001_1003delGCC (p.Ser334del) of the LMNA gene was found. The patient presented with overlapping syndrome of heart failure secondary to dilated cardiomyopathy, limb-girdle dystrophy and partial lipodystrophy. Endomyocardial biopsy revealed strong up-regulation of HLA classes I and II antigens on microvessels and induction of the class I antigens on cardiomyocytes. On muscle biopsy, a wide range of fiber sizes and small clusters of inflammatory infiltrations were found. In the rapid progression of heart failure with arrhythmias or conduction defect, accompanied with muscle atrophy and lipodystrophy, the genetic disease should be taken into consideration. In addition, undefined inflammatory response and fibrosis in the heart or skeletal muscle might further justify screening of the lamin A/C gene.

KEYWORDS: Cardiomyopathy; LMNA gene; Lamin A/C; Laminopathy; Limb-girdle muscular dystrophy

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