A Case of Novel Lamin A/C Mutation Manifesting as Atypical Progeroid Syndrome and Cardiomyopathy.

Guo X¹, Ling C², Liu Y¹, Zhang X², Zhang S³.

Abstract

Mutations in the gene LMNA cause a wide spectrum of diseases that selectively affect different tissues and organ systems. The clinical features of these disorders can overlap but be generally categorized into 2 groups: cardiomyopathy and neuromuscular disorders; premature aging and lipodystrophy disorders. It is significant for a single patient who harbours the 2 sets of diseases simultaneously. We present a female patient with a unique phenotype including rare atypical progeroid syndrome and dilated cardiomyopathy. Genetic mutation detection in the gene LMNA revealed a novel heterozygous de novo mutation p.Leu59Val located in the first exon of gene LMNA c.175C>CG.

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PMID: 27265359 DOI: 10.1016/j.cjca.2015.11.011

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