A novel mutation in lamin a/c causing familial dilated cardiomyopathy associated with sudden cardiac death.


Abstract

BACKGROUND: Dilated cardiomyopathy (DCM), a cardiac heterogeneous pathology characterized by left ventricular or biventricular dilatation, is a leading cause of heart failure and heart transplantation. The genetic origin of DCM remains unknown in most cases, but >50 genes have been associated with DCM. We sought to identify the genetic implication and perform a genetic analysis in a Spanish family affected by DCM and sudden cardiac death.

METHODS AND RESULTS: Clinical assessment and genetic screening were performed in the index case as well as family members. Of all relatives clinically assessed, nine patients showed clinical symptoms related to the pathology. Genetic screening identified 20 family members who carried a novel mutation in LMNA (c.871 G>A, p.E291K). Family segregation analysis indicated that all clinically affected patients carried this novel mutation. Clinical assessment of genetic carriers showed that electrical dysfunction was present previous to mechanical and structural abnormalities.

CONCLUSIONS: Our results report a novel pathogenic mutation associated with DCM, supporting the benefits of comprehensive genetic studies of families affected by this pathology.

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KEYWORDS: Dilated cardiomyopathy; lamin A/C; novel mutation; sudden cardiac death

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