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Case reports of a c.475G>T, p.E159* lamin A/C mutation with a family history of conduction disorder, dilated cardiomyopathy and sudden cardiac death.

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Abstract

BACKGROUND: Patients with some mutations in the lamin A/C (LMNA) gene are characterized by the presence of dilated cardiomyopathy (DCM), conduction abnormalities, ventricular tachyarrhythmias (VT), and sudden cardiac death (SCD). Various clinical features have been observed among patients who have the same LMNA mutation. Here, we show a family with cardiac laminopathy with a c.475G > T, p.E159* LMNA mutation, and a family history of conduction disorder,

DCM, VT, and SCD.

CASE PRESENTATION: A proband (female) with atrial fibrillation and bradycardia was implanted with a pacemaker in her fifties. Twenty years later, she experienced a loss of consciousness due to polymorphic VT. She had a serious family history; her mother and elder sister died suddenly in their fifties and sixties, respectively, and her nephew and son were diagnosed as having DCM. Genetic screening of the proband, her son, and nephew identified a nonsense mutation (c.475G > T, p.E159*) in the LMNA gene. Although the proband's left ventricular ejection fraction remained relatively preserved, her son and nephew's left ventricular ejection fraction were reduced, resulting in cardiac resynchronization therapy by implantation of a defibrillator.

CONCLUSIONS: In this family with cardiac laminopathy with a c.475G > T, p.E159* LMNA mutation, DCM, SCD, and malignant VT occurred. Clinical manifestation of various atrial and ventricular arrhythmias and heart failure with reduced ejection fraction occurred in an age-dependent manner in all family members who had the nonsense mutation. It appears highly likely that the E159* LMNA mutation is related to various cardiac problems in the family of the current report.

KEYWORDS: Case report; Dilated cardiomyopathy; Lamin A/C; Sudden cardiac death; c.475G > T; p.E159*

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