



Pathological Features of Lamin Cardiomyopathy

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A 57-year-old Japanese woman, who had been treated for 8 years as atrial fibrillation (AF) with bradycardia, was admitted because of syncope. Her mother had died suddenly 2 years after implantation of a permanent pacemaker (PPM) and her two elder sisters had also received PPMs for AF with complete atrioventricular block. Electrocardiography of the proband showed AF and an escape rhythm with a heart rate of 42 beats/min. Echocardiography exhibited a mildly enlarged left atrium and ventricle, with mild hypokinesis of the left ventricle (ejection fraction, 51%). Coronary angiography was normal. Histopathology of the endomyocardial biopsy samples from the right ventricle of the proband demonstrated misshapen myocyte nuclei (Figure A), and immunohistochemical staining for anti-lamin A/C antibodies (Cell Signaling Technology, P02545) showed weaker positive reactions in the nuclei of the myocytes (Figure B). Electron microscopic examination showed an intricately indented nuclear

membrane of myocytes (Figure C), and disruption of the nuclear membrane (Figure D). Genetic analysis revealed a heterozygous splicing mutation (IVS1+1G>A) of the lamin A/C gene (*LMNA*) in both the proband and her 26-year-old son, who recorded a normal ECG; the proband had a PPM implanted. Clinicopathological decision, as well as histopathological findings, can lead to the possibility of lamin A/C cardiomyopathy.¹

Disclosures

K.M. is a member of *Circulation Journal* Editorial Team. The other authors have no conflicts of interest to declare.

Reference

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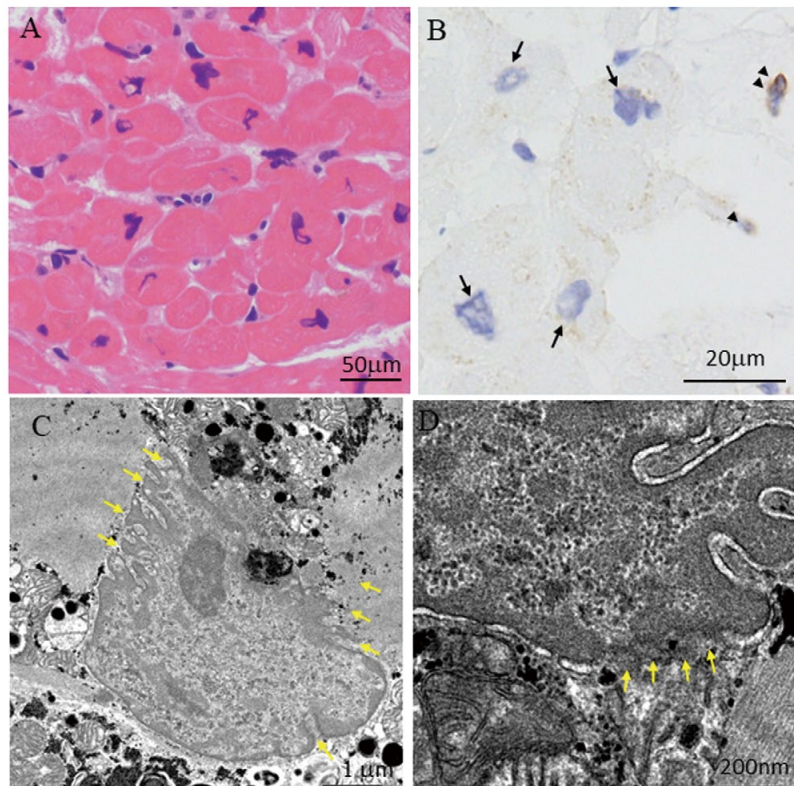


Figure. (A) Histopathologically, the myocytes show misshapen nuclei (H&E). (B) Immunohistochemically, lamin A/C shows a weaker reaction in the nuclei of myocytes (arrows) than in the nuclei of interstitial cells (arrow heads). Electron microscopy shows the intricately indented nuclear membrane of a myocyte (C, arrows) and disruption of the nuclear membrane (D, arrows).