Laminopathies: a Pandora's box of heart failure, bradyarrhythmias and sudden death.

[Article in English, Portuguese]

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Abstract

INTRODUCTION: The LMNA gene encodes a group of proteins that have an important structural and functional role in the cell nucleus. Mutations in this gene have been found in 6% of all forms of dilated cardiomyopathy and in up to 33% of those with conduction system disturbances.

AIMS AND METHODS: Using a case report as an example, we performed a review of the literature on the pathophysiological mechanisms, clinical manifestations, risk stratification and treatment options of cardiac involvement in laminopathies.

CASE REPORT: We present the case of a 46-year-old man, whose ECG showed bizarre voltage criteria for left ventricular hypertrophy and first-degree atrioventricular block, a dilated left ventricle with mildly impaired global systolic function and non-sustained ventricular tachycardia on Holter monitoring, and with a family history of sudden death. Genetic testing identified an LMNA mutation. No ventricular arrhythmias were induced during electrophysiological study. The patient is under close clinical and echocardiographic monitoring and an event loop recorder has been implanted.

DISCUSSION: Phenotypically, myocardial involvement in laminopathies is indistinguishable from other forms of idiopathic dilated cardiomyopathy. Ventricular arrhythmias are common, but the best method for sudden death risk stratification has yet to be established. The few studies that have been performed, with a very limited number of patients, show that factors associated with an unfavorable prognosis are ejection fraction <45%, non-sustained ventricular tachycardia, male gender and any form of atrioventricular block. Given the lack of evidence, indications for an implantable cardioverter-defibrillator for primary prevention in this context are the same as...
conventional indications for other forms of idiopathic dilated cardiomyopathy.

**CONCLUSIONS:** Cardiac involvement as a consequence of LMNA mutations generally has a more aggressive natural history than other forms of non-ischemic dilated cardiomyopathy. A high index of suspicion and prompt referral for genetic testing are essential for appropriate therapeutic management.

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**KEYWORDS:** Dilated cardiomiopathy; LMNA gene mutations; Laminopathy; Laminopatia; Miocardiopatia dilatada; Mutações no gene LMNA

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