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Myocardial fibrosis as a early cardiac marker of disease in patients with lamin A/C mutations

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Objective

to assess myocardial fibrosis in lamin A/C mutations (LM) carriers using constrast-enhanced cardiac magnetic resonance (CMR).

Background

LM carriers usually develop a dilated cardiomyopathy (DCM) phenotype in association with atrio-ventricular blocks and/or ventricular arrhythmias. However, sudden cardiac death may be the first clinical manifestation. Hence early detection of myocardial abnormalities before overt left ventricular (LV) dysfunction is warranted for a better risk stratification.

Methods

Seventeen paucisymptomatic LM carries (age 41±16 years, 9 male), 14 paucisymptomatic DCM patients (age 59±11 years, 8 male, NYHA I-II) with mild LV dysfunction (ejection-fraction 40%-55%) and 12 healthy controls (age 46±10 years, 9 male) underwent complete clinical, echocardiographic, biohumoral and contrast-enhanced CMR assessment. Cine CMR was used to derive LV volumes, mass and function, and post-contrast CMR to detect late gadolinium enhancement (LGE) as an index of gross fibrosis. Eight LM patients, all DCM patients and all normal controls underwent pre- and post-contrast myocardial T1 mapping with gadolinium partition coefficient calculation, as an index of interstitial fibrosis.

Results

LM carriers presented a similar LV end-diastolic volume (85±17 ml/m2, p=NS) and slightly reduced ejection-fraction (61±6% p<0.001) than controls (83±19 ml/m2, 67±6%), while in DCM patients LV end-diastolic volume (102±17 ml/m2) and ejection-fraction (49±5%) were worse than both controls and LM carriers (p<0.001). Moreover, DCM patients presented worse diastolic dysfunction (9±3 vs 7±1, p<0.01), NT-proBNP plasma levels (438±416 vs 108±139 ng/l, p<0.01) and Doppler-estimated pulmonary pressure (33±7 vs 30±5 mmHg, p<0.01) than LM carriers. Six (43%) DCM patients presented DE, representing 9±3% of LV mass; similarly, seven (42%) LM carriers had DE, but with a larger extension (11±5% of LV mass, p<0.03). In LM carries DE distribution was patchy (2 pts) or mid-wall (5 pt), similarly to DCM patients (patchy in 1 pt, mid-wall in 4 pts, subendocardial in 1 pt). GPC reached a steady state 5 minutes after gadolinium injection. In DCM patients GPC was slightly higher than controls (0.42±0.05 vs 0.38±0.06, p=0.04), while in LM carriers it was much higher (0.45±0.07) than controls (p<0.01). Even excluding hyperenhanced myocardium, in LM carriers GPC remained higher than controls, while in DCM patients only slightly higher than controls.

Conclusions

Myocardial fibrosis occurs early in LM carrier before LV dilatation and dysfunction and may represent an early phenotypic expression of cardiac involvement.

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