Arrhythmogenic left ventricular cardiomyopathy and dilated cardiomyopathy: genotype-phenotype correlations

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Topic(s):
Cardiac Magnetic Resonance: Myocardium

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Background: The phenotype distinction between arrhythmogenic cardiomyopathy affecting the left ventricle (ALVC) and dilated cardiomyopathy (DCM) is challenging, due to apparent clinical and imaging overlap.

Objectives: To compare arrhythmic, structural, functional and tissue characterization phenotypes of genetically-defined ALVC and DCM.

Methods: Eighty patients with pathogenic ALVC-associated mutations (desmoplakin n=25, filamin C n=7) and DCM-associated mutations (titin n=30, lamin A/C n=12, bcl2-associated thanogene 3 n=3 and RNA binding motif protein 20 n=3) were phenotyped by clinical, electrocardiographic and cardiovascular magnetic resonance (CMR) features.

Results: There were no significant differences in age, sex, symptoms, baseline electrocardiography, arrhythmia burden or ventricular volumes between the two groups. Premature ventricular contractions and ventricular tachycardias were more frequently polymorphic in ALVC genotypes (37.5 vs 14.6% in DCM genotypes, p=0.030). Absolute LV global longitudinal strain was lower in DCM genotypes (median -11.3 vs -14.1% in ALVC genotypes, p=0.024) but LV regional wall motion abnormalities were more common in ALVC genotypes (46.9 vs 4.2% in DCM genotypes, p<0.001). Sub-epicardial late gadolinium enhancement of LV myocardium with a ring-like pattern (at least 3 contiguous segments in the same short axis slice) was observed in 78.1% of ALVC genotypes but was absent in DCM genotypes (p<0.001).

Conclusion: We showed that when ALVC and DCM are defined genetically, DCM has more impaired global longitudinal strain (despite same ejection fraction and dilatation), but that ALVC is more heterogeneous with more regionality in LV impairment and with more polymorphic ventricular arrhythmia. The most characteristic difference however is that ALVC frequently has ring-like late gadolinium enhancement not found in genetic DCM. This should be considered in future diagnostic criteria for ALVC.
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