Imaging: Myocardial Disease

Incremental value of cardiovascular magnetic resonance imaging in family screening for hypertrophic cardiomyopathy

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Genetic testing in relatives of hypertrophic cardiomyopathy (HCM) patients can lead to early identification of carriers of pathogenic DNA variants (G+), before onset of left ventricular hypertrophy (LVH). Repeated evaluation by electrocardiography (ECG) and transthoracic echocardiography (TTE) is recommended to detect HCM during follow-up. Cardiovascular magnetic resonance (CMR) imaging has become valuable in the work-up of HCM, although its role in G+ subjects has not been extensively evaluated. In this study, we investigated the value of CMR in the G+/LVH- population.

We included 55 G+ subjects who underwent CMR in addition to ECG and TTE, with a maximal wall thickness (MWT) <15mm on TTE. The CMR imaging protocol consisted at least of steady state free procession imaging and 2-dimensional late gadolinium enhancement (LGE) images. ECGs were considered abnormal in case of pathologic Q waves, T wave inversion or signs of LVH (by voltage criteria including Sokolow-Lyon and a Romhilt-Estes score ≥4). TTEs were abnormal in case of LVH (defined as MWT≥10mm). For both modalities, the diagnosis of HCM was based on a MWT≥13mm. The yield of CMR relative to ECG/TTE was assessed by comparing the proportion of HCM diagnoses and the presence of other phenotypic features. Forward step logistic regression was used to assess whether the presence of TTE/ECG abnormalities could predict reclassifications or abnormalities (crypts and LGE) on CMR.

An overview of ECG/TTE and CMR findings is shown in the Figure. Two of 16 (13%) subjects diagnosed with HCM on TTE were reclassified as having no HCM on CMR, and 8 of 39 (21%) subjects without HCM on TTE were reclassified as HCM on CMR. These 8 subjects had a mean MWT of 15.4 ± 2.6 mm on CMR and a mean MWT difference of 4.5 ± 2.9 mm (range 1.7-9.4) compared to TTE, which in 3 cases was explained by a hook-shaped thickening of the basal anterior wall in the 2 chamber view, not visible on TTE. Compared to subjects without HCM on both modalities, the reclassified group had a significantly higher QRS duration (104 ± 14 vs 93 ± 11 ms, p = 0.03) and anterior mitral valve leaflet length (30 ± 4 vs 26 ± 3 mm, p = 0.01). Of the 13 subjects with normal ECG/TTE results, none were reclassified as HCM using CMR.

The proportion of additional CMR abnormalities was large in subjects with and without abnormal ECG/TTE results (57% vs 38%, p = 0.24). Subjects with poor TTE image quality were equally likely to be reclassified compared to those with sufficient image quality (10% vs 24%, p = 0.19). Logistic regression demonstrated that the presence of TTE/ECG abnormalities (odds ratio [OR] 8.7 [1.3-59.0], p = 0.03) and age (OR 1.1 [1.0-1.2], p < 0.01) independently predicted reclassifications or presence of abnormalities using CMR.

Additional CMR imaging reclassifies 18% of subjects. Subjects with normal ECG and TTE results are not diagnosed as HCM on CMR, but the prevalence of HCM-related abnormalities on CMR was high in subjects with and without ECG/TTE abnormalities.

Abstract Figure. Diagnostic approach and CMR findings