Case Report

Hypertrophic cardiomyopathy in Noonan Syndrome closely mimics familial hypertrophic cardiomyopathy due to sarcomeric mutations

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Abstract

A 27 year old female with Noonan syndrome and hypertrophic cardiomyopathy underwent cardiovascular magnetic resonance imaging. These images showed asymmetrical septal hypertrophy with maximal left ventricular end-diastolic wall thickness of 25 mm. Following administration of gadolinium, areas of hyperenhancement were seen in the anterior, anteroseptal and lateral walls. This is the first report of focal gadolinium hyperenhancement in hypertrophic cardiomyopathy due to Noonan syndrome and suggests that myocardial fibrosis can be imaged by MR hyperenhancement as seen previously in sarcomeric hypertrophic cardiomyopathy.

A 27 year old female with Noonan syndrome and hypertrophic cardiomyopathy underwent a cardiovascular magnetic resonance (CMR) scan as part of a research study. As a child she had had an open pulmonary valvotomy and a secundum atrial septal defect closure. She was symptomatically well to follow up with no chest pain or shortness of breath.

CMR was performed using a 1.5 T Sonata Siemens System (Siemens Medical Solutions, Erlangen, Germany) with body coil and phased array surface coil, prospective electrocardiographic gating and the patient in the supine position. After piloting, steady state free precession cine images (TE/TR 1.5/3.0 ms, flip angle 60°, slice thickness 7 mm with 3 mm interslice gap, in-plane resolution 1.5×1.5mm\(^2\), temporal resolution 45 ms, breath-hold duration 14–17 heartbeats per breath-hold) were acquired in the horizontal and vertical long axis views during breath-hold in end-expiration. Image analysis was performed with Argus Software (Version 2002B, Siemens Medical Solutions).

These images showed asymmetrical septal hypertrophy (Figure 1A and B) with increased LV mass index (112 g/m\(^2\), normal <72 g/m\(^2\)) with a maximal end-diastolic LV wall thickness of 25 mm in the basal anterior myocardium. The left ventricle was hyperdynamic (ejection fraction 80%, normal range 56–79%) and there was no residual shunt and no resting left ventricular outflow tract gradient.
Images were then acquired following the injection of 0.1 mmol/kg body weight of gadolinium-DPTA. Patchy, signal-intense areas were seen in the anterior, anteroseptal and lateral walls (Figure 1C and D, white arrows). In this patient, these areas represented 9.4% of left ventricular mass.

Discussion

The hypertrophic cardiomyopathy seen in Noonan Syndrome is the closest cardiac phenocopy of familial hypertrophic cardiomyopathy caused by sarcomeric mutations and includes the typical and extensive myocardial disarray [1]. Previous studies have demonstrated focal gadolinium enhancement in sarcomeric hypertrophic cardiomyopathy [2, 3] which correlates with fibrosis on histological specimens [4]. The degree of hyperenhancement has recently been correlated to ventricular dilation and markers of sudden death in a blinded prospective study of sixty-three patients with hypertrophic cardiomyopathy [5]. This cardiac MRI case report underlines the anatomical and pathophysiological parallels between HCM phenocopies. It is the first report of gadolinium hyperenhancement in hypertrophic cardiomyopathy due to Noonan syndrome.

References