CASE REPORT

Diagnosis of apical hypertrophic cardiomyopathy using magnetic resonance imaging

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Abstract
Apical hypertrophic cardiomyopathy is an uncommon variant of non-obstructive hypertrophic cardiomyopathy with low prevalence outside East Asia. A case is reported of a non-Asian (European) 51 year old man with characteristic ECG and morphological changes of apical hypertrophic cardiomyopathy. Although the patient underwent catheterisation three years previously because of suggested coronary ischaemic heart disease, apical hypertrophic cardiomyopathy was not diagnosed. More recently, a regional wall motion abnormality was noticed at the apex on echocardiography. To rule out an ischaemic injury a stress perfusion scintigraphy was performed; no perfusion defect was present but an apical tracer enhancement was noted. Further evaluation by magnetic resonance imaging revealed the pathognomonic "ace of spades" configuration of the left ventricle with systolic obliteration of the apical region, which led to the diagnosis of apical hypertrophic cardiomyopathy.
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Keywords: apical hypertrophic cardiomyopathy; magnetic resonance imaging

Magnetic resonance imaging (MRI) is the most appropriate non-invasive test available to assess the extent and severity of left ventricular hypertrophy especially in patients with poor echogenicity. Diagnosis of apical hypertrophic cardiomyopathy is important because it carries a more favourable prognosis than other forms of hypertrophic cardiomyopathy.

Case report
A 51 year old European (white) man complained initially of atypical chest pain in 1996. He had never smoked and had no other cardiovascular risk factors. An ECG showed giant negative T waves in the precordial leads (V2–6) which progressed during exercise. Echocardiography did not reveal any further information owing to limited image quality. The patient underwent left ventricular catheterisation, which excluded coronary artery disease. Left ventricular hypertrophy with normal systolic function (ejection fraction 72%) was diagnosed. The patient was asymptomatic in the following years.

In January 1999 the patient felt a slight progressive shortness of breath at exercise (New York Heart Association functional class I). ECG at rest showed similar abnormalities to previously (fig 1). Transthoracic echocardiography revealed hypokinesia of the apex, but no further information was available owing to limited image quality.

The patient was referred to our department for a myocardial perfusion scintigraphy. Gated SPECT perfusion study with thallium-201 at rest and under stress showed a regional tracer enhancement of the apical region (fig 2). During exercise the ECG showed progression of T wave inversion and a significant descending ST segment lowering in the precordial leads. Despite normal perfusion pattern, the gated images indicated a hypodynamic movement of the apex.

Because of the discrepancy in impaired wall motion and normal perfusion in the apical region, an ECG gated cinemagnetic resonance examination was performed. Magnetic resonance data showed localised hypertrophy of the apical region with a spade-like configuration of the left ventricle at end diastole, and an obliteration of the distal left ventricular cavity at end systole (figs 3 and 4). Basal cardiac regions showed a normal wall thickness; systolic function was also normal (ejection fraction 68%).
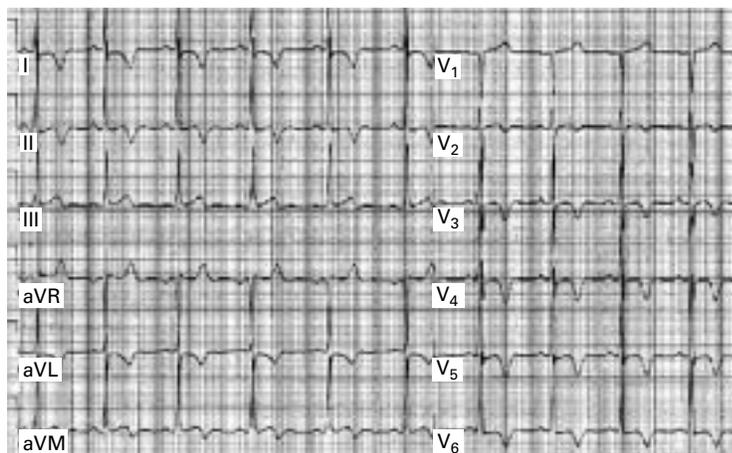

Figure 1   ECG at rest with negative T waves in the precordial leads and a positive Sokolow-Lyon index (4.1 mV) as signs of left ventricular hypertrophy.
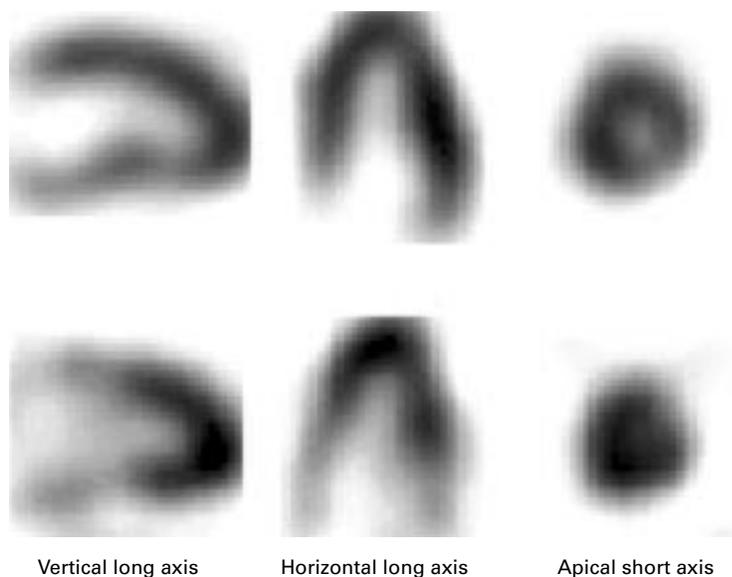
Vertical long axis   Horizontal long axis   Apical short axis

Figure 2   Gated SPECT images showing a rich perfusion pattern in the apical region at rest (top) and under stress (bottom).
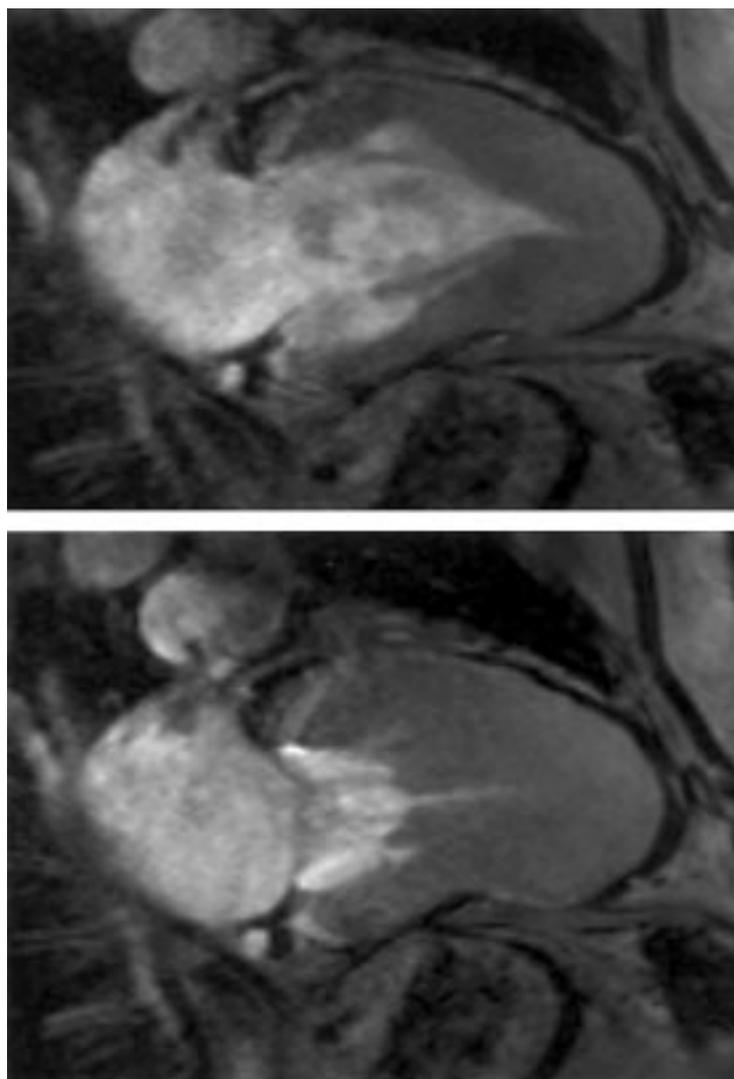

Figure 3   Spade-like configuration of the left ventricle at end diastole (top) in two chamber magnetic resonance images with obliteration of the distal cavity at end systole (bottom) (fast gradient echo technique).

Considering the MRI results in combination with the ECG changes, the patient was diagnosed with apical hypertrophic cardiomyopathy. Dyspnoea was assessed as a sign of disturbed diastolic relaxation.

Discussion

Hypertrophic cardiomyopathy is a primary disease of cardiac muscle with a wide variety of clinical and morphological expression. Left ventricular hypertrophy can be diffuse or limited to specific regions of the left ventricle.

The first case of apically localised hypertrophic cardiomyopathy was described in Japan in 1976.[1] Since then, this issue has received considerable attention and several reports have been published in and outside Japan.[2–4] It seems that apical hypertrophic cardiomyopathy presents variations in its manifestation among many Japanese patients and most cases described in studies from outside Japan. The reason is unknown, but genetic, racial or even environmental factors could be responsible.

Global prevalence of hypertrophic cardiomyopathy is between 0.02 and 0.2%. In Japan, the apical variant makes up approximately 25% of all hypertrophic cardiomyopathy cases, the prevalence outside Japan is rather low (1–2% of all hypertrophic cardiomyopathy).[3]

Histological examination of apical hypertrophic cardiomyopathy shows a severe disorganisation of myocardial muscle fibres similar to other forms of hypertrophic cardiomyopathy.[4] The pathogenesis of apical hypertrophic cardiomyopathy is not yet clear, but a familial prevalence is likely, similar to patients with hypertrophic cardiomyopathy. While an autosomal dominant type is usually found in patients with hypertrophic cardiomyopathy, some investigators have supposed a sex linked recessive inheritance.[2] An association with HLA-DR2 antigen in patients with apical hypertrophic cardiomyopathy was recently described in Japan.[5] Other investigators consider a secondary genesis as the underlying pathogenetic mechanism—that is, hypertension or heavy physical exercise.[6]

Pathophysiological studies in patients with apical hypertrophic cardiomyopathy revealed an impaired left ventricular diastolic function with a decreased rate of early diastolic filling, and increased contribution to the atrial contraction, as can be seen in other forms of left ventricular hypertrophy. In contrast, systolic function seems to be normal or even hyperdynamic.[6] Depending on the severity of reduced relaxation, intraventricular end diastolic and filling pressures could be raised.

Atypical chest pain is the most frequent symptom in patients with apical hypertrophic cardiomyopathy. Typical angina may also occur because of diminished vasodilatatory reserve, similar to patients with asymmetrical septal or secondary hypertrophy owing to arterial hypertension or aortic valve stenosis. Other common clinical findings are dyspnoea, palpitations, reduced exercise tolerance, and increased fatigue. As clinical symptoms are non-specific, the diagnosis of apical hypertrophic cardiomyopathy is primarily made from ECG
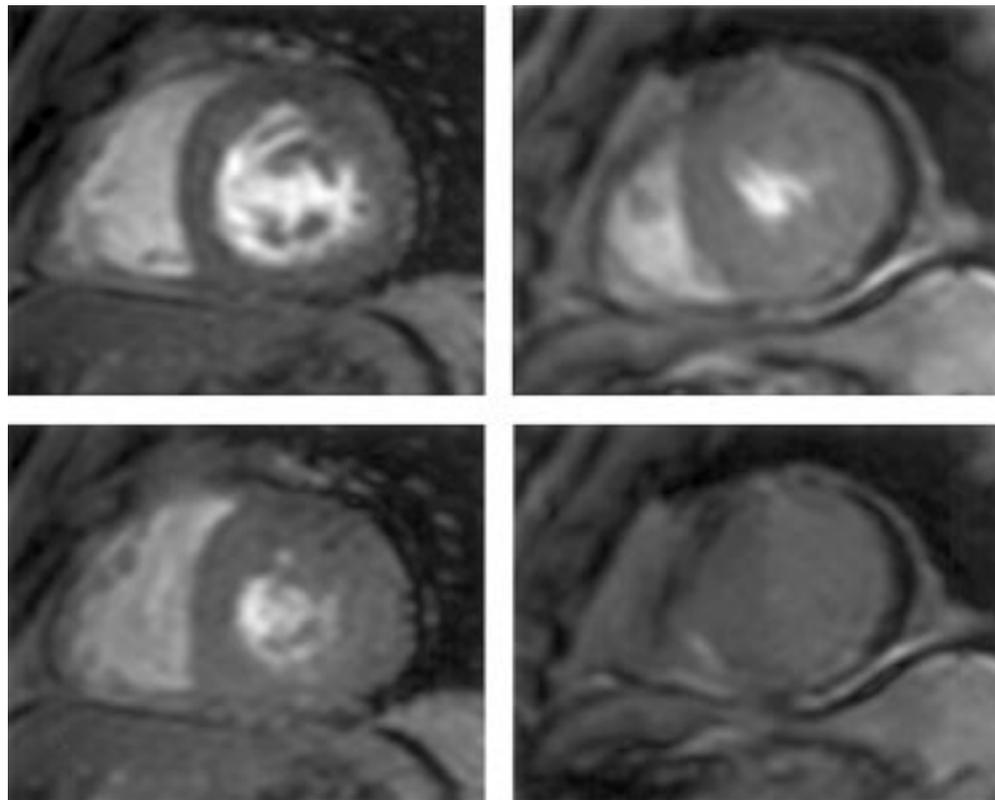
Figure 4   Basal short axis magnetic resonance images (left) show normal diastolic wall thickness at end diastole (above) and end systole (below). Distal short axis slices (right) reveal the apical hypertrophy at end diastole (above) with obstruction of the cavity at end systole (below) (fast gradient echo technique).

changes and specific morphological criteria. While morphology was assessed by angiography in early cases, later studies focused on non-invasive assessment with echocardiography.

Typical ECG changes are signs of left ventricular hypertrophy such as pronounced negative T waves (> 1 mV) in the precordial leads and a positive Sokolow-Lyon index.[2] In some non-Japanese studies, T wave negativity was often less pronounced and not necessarily localised to the precordial leads.[4]

Cineangiography in the right anterior oblique (RAO) projection shows a spade-like ("ace of spades") configuration of the left ventricle at end diastole as expression of apical hypertrophy, with obliteration of the distal cavity at end systole.[2] This spade-like configuration is less often seen in patients from outside Japan, because hypertrophy in these patients is often not strictly localised to the apex.[6] Other common invasive findings are an increased ratio ($\geq$ 1.5) between apical wall thickness and thickness of the midportion of the free anterior wall, mild mitral regurgitation, raised left ventricular end diastolic pressures, and a prominent A wave in pulmonary capillary wedge pressure tracings.[2]

Transthoracic echocardiographic findings vary with the extension of hypertrophy. Significant narrowing towards the apex of the left ventricle without abnormalities in other segments of the heart is usually found in apical hypertrophy.[3] This can result in a significant obliteration of the distal left ventricular cavity, which can be misinterpreted as wall motion abnormality, especially when assessment of the endocardial border is difficult, as in our patient.

Blood pool scintigraphy at rest usually does not reveal any abnormalities, but an increase of left ventricular ejection fraction during stress is often found as a result of hyperdynamic systolic contraction.[6]

Myocardial perfusion SPECT at rest and during stress is usually normal. Sometimes, reversible perfusion defects of the apical region are seen without significant coronary artery disease owing to a disproportion of regional wall thickness and vascular supply.[6] Despite normal perfusion, wall motion abnormalities of the apex can appear in gated acquisition as a result of distal cavity obstruction as observed in our patient.

ECG gated MRI provides reliable assessment of morphological as well as functional information of the myocardium. Compared with echocardiography, MRI allows a better assessment of left ventricular endocardial border, myocardial mass, and global and regional wall motion. MRI is therefore a valuable technique for studying patients with hypertrophic cardiomyopathies. Imaging in standard planes (transverse, sagittal, coronal) is often insufficient for the assessment of regional hypertrophy. Section planes, comparable with those used in cross sectional echocardiography, allow better judgment of localisation and extension of hypertrophy.[6] There are few reports concerning the evaluation of apical hypertrophic cardiomyopathy by MRI.[6–9]

In MRI, the typical spade-like configuration can be demonstrated on left ventricular long

axis images similar to the RAO projection in angiography. The configuration in this projection is caused by hypertrophied myocardium of the apical anterior and the apical posterior wall. Patients with the typical spade-like configuration show a circumferential hypertrophy of the entire apex in short axis MRI.

A subtype of apical hypertrophic cardiomyopathy was previously identified on magnetic resonance short axis images with asymmetric apical hypertrophy.[7] The distribution of hypertrophied myocardium in this subtype is frequently proved to be confined to the apical lateral wall, which therefore cannot be evaluated on long axis images or RAO projections in angiography. Thus, this subtype was called non-spade apical hypertrophic cardiomyopathy. Although the features between the two configurations are quite different, giant negative T waves are common in both.

Recently, a long term study showed that patients with non-spade apical hypertrophic cardiomyopathy developed the typical spade-like configuration after some years.[8] Thus, the non-spade type is possibly an early stage of apical hypertrophic cardiomyopathy.

In our patient, the extension of hypertrophy of the apical region with typical spade-like configuration was clearly seen on long axis MRI. Short axis images showed a circumferential hypertrophy of the entire apex.

There is not much known about the treatment of apical hypertrophic cardiomyopathy. Most patients are asymptomatic and therefore probably do not require specific treatment. Treatment of diastolic dysfunction is difficult. Although calcium channel blockers showed a beneficial effect in patients with hypertrophic cardiomyopathy, results are rather disappointing in apical hypertrophic cardiomyopathy. In patients with typical angina, some authors recommend the use of β blockers or calcium channel blockers to diminish myocardial oxygen consumption.[6] β Blocker or frequence lowering calcium channel blockers can be used if arrhythmias or palpitations are present.

Considering the reports published thus far, prognosis of apical hypertrophic cardiomyopathy appears to be favourable in most patients. In a few rare cases, ventricular tachycardia occurred and sudden death has been reported.[4] Another rare complication is myocardial infarction of the apex, despite normal coronary angiographic findings, which is explained by a disproportion of vascular supply and myocardial wall thickness.[10]

CONCLUSION

Apical hypertrophic cardiomyopathy is a rare form of hypertrophic cardiomyopathy with regional difference in expression. As symptoms are not specific, the diagnosis is based on ECG changes and morphological criteria. While morphological assessment was initially based on invasive procedures, the diagnosis can be made non-invasively. MRI in different planes is a valuable non-invasive technique to assess regional morphology and myocardial function, especially in cases where echocardiography cannot provide clear information. In patients with severe ECG changes, spade and non-spade apical hypertrophic cardiomyopathy should be considered, and one should bear in mind that in the latter form short axis images should be obtained for diagnosis.

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