

Apical hypertrophic cardiomyopathy in an adult (RCD code: III-2A)

Kamil Bugała*¹, Wojciech Płazak², Małgorzata Konieczńska¹, Piotr Podolec²

¹ Department for Diagnosis, John Paul II Hospital, Krakow, Poland; ² Department of Cardiac and Vascular Diseases, John Paul II Hospital, Jagiellonian University Medical College, John Paul II Hospital, Krakow, Poland

Abstract

Apical hypertrophic cardiomyopathy (AHC) is a less common type of hypertrophic cardiomyopathy (HCM). It is characterized by typical changes in ECG (deeply inverted T waves that can mimic acute coronary syndrome), echocardiography (hypertrophy of apical segments) and ventriculography (spade-like shaped left ventricular cavity). Magnetic resonance is considered as a reference method for AHC diagnosis. We present a long follow-up period of a patient with this form of HCM. The management of patients with HCM is directed at decreasing left ventricle outflow tract narrowing (if present), decreasing of intraventricular gradient (if present), heart failure treatment and prevention of sudden cardiac death. In general treatment adequate hydration is essential and diuretics should be used with caution to prevent the increase of intraventricular gradient. JRC D 2016; 2 (6): 185–188

Key words: congenital heart disease, heart failure, rare disease, ventriculography, echocardiography, spade-like shape

Case presentation

An adult male, aged 50 years was referred to our Clinic for the first time in 1999 with a suspicion of an acute coronary syndrome. His medical history included hypertension (for about 15 years), blood lipid disorders and no sudden cardiac death in his family history. He complained of a decrease of exercise tolerance with intermittent chest pain at rest and after exertion. Electrocardiogram (ECG) showed sinus rhythm 78 bpm, deep negative T waves with ST segment depressions in leads I, II, aVL, V2-V6 (Figure 1). Coronary angiography showed no significant atheromatous lesions and ventriculography revealed massive left ventricular (LV) hypertrophy including apex and apical segments with total systolic obliteration of the LV cavity from apex to mid segments, without intraventricular gradient (Figure 2). Transthoracic echocardiography (TTE) showed muscle hypertrophy as described above, impaired LV relaxation with normal systolic function (EF 65%) and non-significant left atrium (LA) enlargement. No valvular dysfunction was found. The diagnosis of apical hypertrophic cardiomyopathy (AHC) was established and therapy with beta-blocker (metoprolol), *dihydropyridine* calcium blocker (amlodipine) and angiotensin converting enzyme inhibitor (quinapril) was initiated. Genetic

testing was not performed. The patient was discharged home with recommendation of routine follow-up visits in outpatient clinic.

In 2001 the patient was admitted to the Clinic again for cardiological evaluation due to uncharacteristic chest pain episodes. ECG changes did not differ from those observed on prior tracings. Taking into consideration the risk factors of coronary artery disease (CAD), coronary computed tomography angiography was performed. It showed no significant obstructive CAD. Chest radiography revealed thoracic scoliosis. Cardiac magnetic resonance (CMR) showed severe hypertrophy of apical segments with apical septal segment thickness of 22 mm and lateral apical segment of 31 mm (Figure 3). LV size and function were normal and LV mass was assessed as 343g. On delayed enhancement imaging, patchy areas of enhancement in the hypertrophic apical segments were observed – the image corresponded with microvascular dysfunction. He was discharged home after pharmacotherapy modification, that included increasing the dose of amlodipine from 5 mg to 15 mg once a day and changing the type of beta-blocker from metoprolol 50 mg twice a day to bisoprolol 5 mg once a day.

Follow-up CMR performed in July 2003 showed obliteration of the LV from mid segments to apex in systole and LV mass similar to the previous assessment. Of note, in comparison to CMR done in

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* Corresponding author: Department for Diagnosis, John Paul II Hospital, Prądnicka str. 80, 31-202 Krakow, Poland; tel. 0048 12 614 25 56; e-mail: kamil.bugala04@gmail.com

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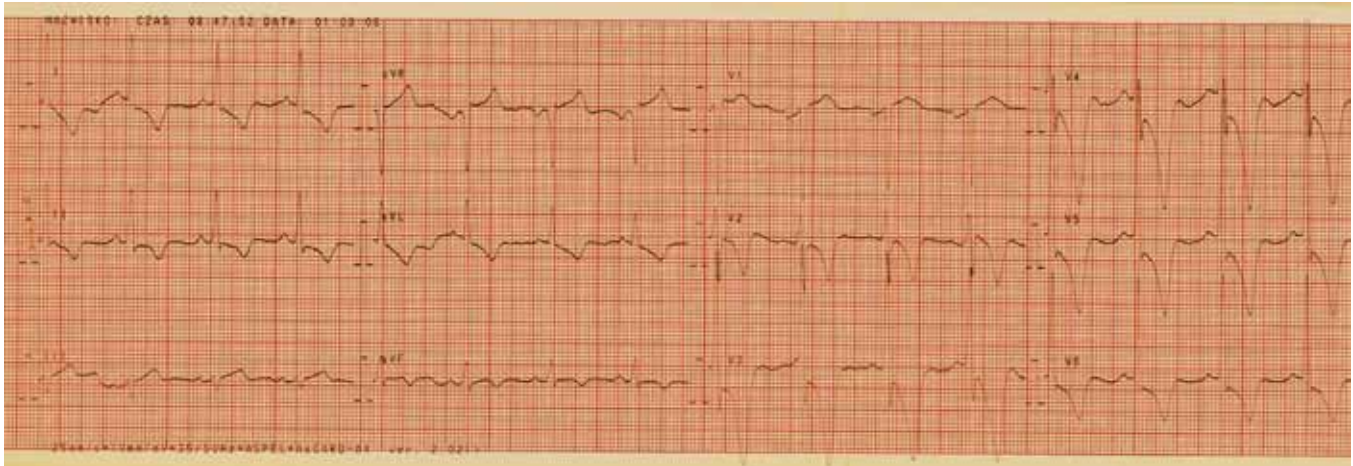


Figure 1. Electrocardiogram. Sinus regular rhythm of ventricular frequency 78 beats per minute, normal axis, QRS complex duration 100ms, deep negative T waves with ST segment depressions in I, II, aVL, V2-V6

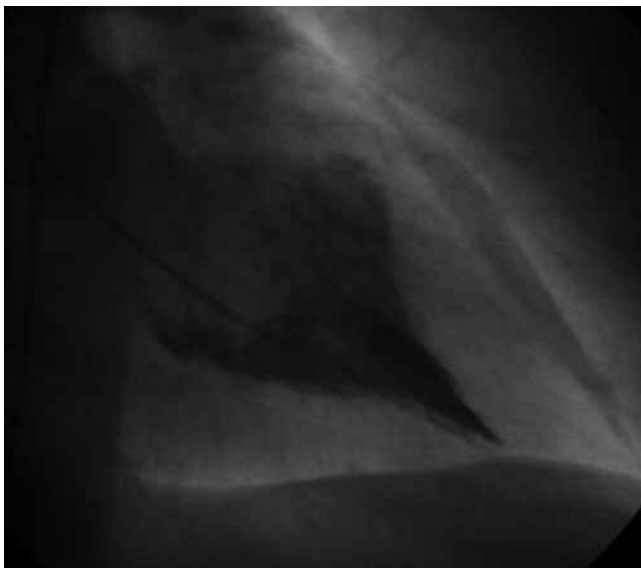


Figure 2. Left ventriculography. Left ventricular (LV) hypertrophy including apex and apical segments with spade-like shaped LV cavity

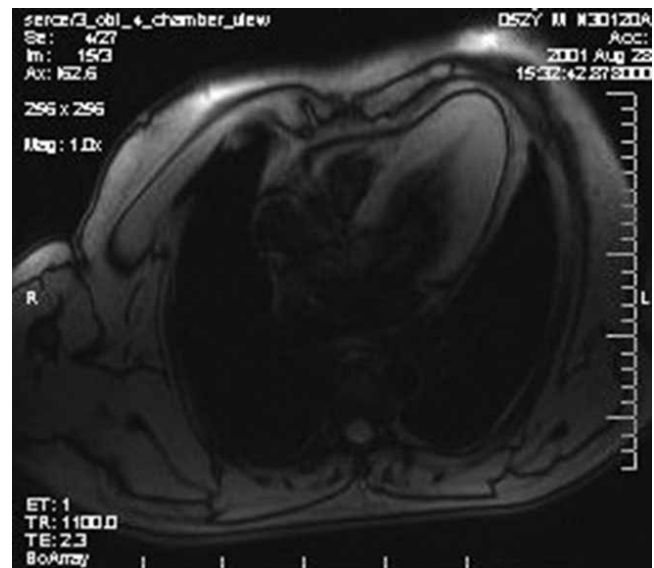


Figure 3. Cardiac magnetic resonance. Four chamber projection showing severe hypertrophy of apical myocardial segments

2001 year, clamping area of the LV was closer to the base of the heart and these findings indicated the ongoing remodeling of LV.

At that time the patient was in general good condition. Routinely seen in outpatient clinic, he reported good tolerance of physical effort (class I by New York Heart Association). However, antithrombotic therapy (warfarin) was started due to recurrent episodes of paroxysmal atrial fibrillation (AF).

In December 2015 he was admitted to the hospital with the symptoms of acute heart failure. A week before admission he reported worsening of dyspnea and the dose of diuretics were increased by general practitioner. This time ECG showed AF with ventricular response rate at 100 bpm, deep negative T waves with ST segment depression in leads I, II and V2-V6. On physical examination loud systolic murmur in Erb's point was present. Laboratory work-up revealed abnormalities in blood lipid profile (LDL cholesterol of 3,09 mmol/l, triglycerides of 1,93mmol/l), mildly el-

evated level of liver transaminases that could be caused by usage of statins (ALAT of 51 U/L [normal <41 U/L] and AspAT of 53 U/L [normal <40 U/L]). Other routine laboratory tests were within normal range. Admission echocardiography showed systolic obliteration of the LV with substantial dynamic intraventricular gradient (90 mmHg, up to 120–150 mmHg after extra ventricular contractions). Significant enlargement of LA was also observed (50x60mm, area 41,5 cm²), apical segment's thickness of LV myocardium was up to 30 mm (Figure 4). LV was normal in size (45/25 mm) and had hyperdynamic systolic function (EF 80%). Features of diastolic dysfunction were noticed without significant valvular dysfunction. This time pharmacological therapy was modified by changing type and increasing the dose of beta-blocker (Metoprolol Succinate) up to maximum tolerated dose 200mg/day. Amlodipine, aldosterone receptor blocker (spironolacton), statin agents, oral anticoagulant (warfarin) was continued. Moreover, metformin was started due to newly diagnosed type 2 diabetes. 24-hour ECG Holter did not

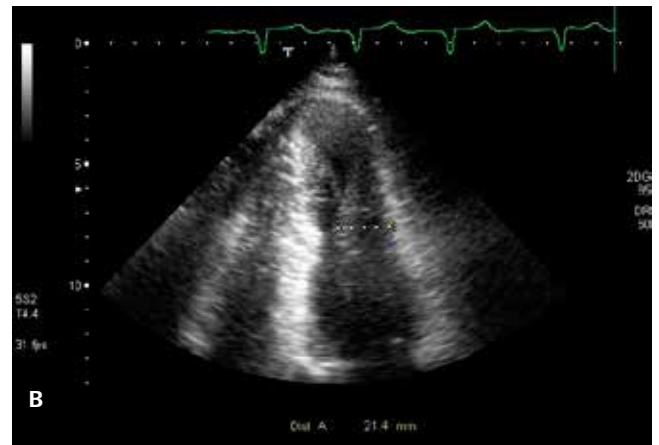


Figure 4. Transthoracic echocardiography. Apical four chamber views. A,B. Massive left ventricular (LV) hypertrophy including apical segments. C. Hypertrophy of mid segments with narrowing of LV lumen

reveal any significant ventricular arrhythmias, but AF was registered during entire monitoring time. Introduced treatment improved the patient's general condition. Control echocardiogram showed reduction of intraventricular gradient to its maximum at 30 mmHg (Figure 5). Individualized risk of sudden cardiac death (SCD) has been calculated to be 2,77% at 5 years, therefore according to the current European Society of Cardiology (ESC) guidelines the decision of ICD implantation was postponed [1]. The patient was discharged home and clinical follow-up visit including echocardiographic assessment was scheduled in next two weeks.

Epidemiology

Hypertrophic cardiomyopathy (HCM) is an inherited heart disease defined by increased LV wall thickness (≥ 15 mm in one or more LV myocardial segments), which cannot be explained by abnormal loading conditions (e.g. arterial hypertension, aortic stenosis) [1]. HCM is one of the most common inherited heart disease observed in 1 in 500 of the general adult population and mostly it is caused by mutations in genes encoding the different components of the sarcomere apparatus [2,3]. The phenotypic expression is variable and one of the variant may present as the AHC. It was first described by Japan researchers in 1976 and it was characterized by deeply inverted T wave in ECG with the demonstration of localized apical hypertrophy in imaging studies [4,5]. The oc-

currence of AHC varies highly in the literature, ranging from 1 to 25% in HCM [6,7]. It is common in Japanese and other Asian populations and less common in non-Asian patients.

Management

The management of patients with HCM is directed at decreasing of LV outflow tract narrowing (if present), decreasing of intraventricular gradient (if present), heart failure treatment, prevention of SCD and treatment of concomitant diseases (e.g. AF which develops in ~20% of patients) [8]. Moreover, screening of family members is suggested. Treatment options for HCM include management with beta-blockers, verapamil and non-pharmacologic management such as, septal myotomy or transcatheter alcohol ablation of the intraventricular septum [9]. Invasive options are indicated when the symptoms become refractory to pharmacological treatment. SCD in high-risk patients may be prevented with the use of implantable cardioverter-defibrillators.

There are five main clinical SCD risk factors [10]:

- extreme LV hypertrophy (>30 mm);
- blunted blood pressure response to exercise in patients younger than 40 years;
- family history of SCD;
- unexplained syncope;
- nonsustained ventricular tachycardia on 24-hour ECG Holter monitoring.

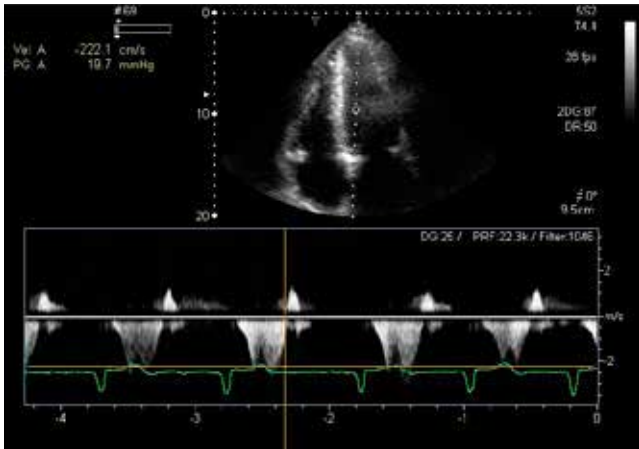


Figure 5. Transthoracic echocardiography. Apical four chamber view. Continuous wave doppler showing intraventricular gradient – study performed after modification of therapy

In 2014 year, a novel clinical risk prediction model was developed and validated for SCD in HCM [11]. This model provides individualized 5-year risk of SCD.

LV mid-cavity obstruction occurs in approximately 10% of patients with HCM [12,13]. The patients with LV mid-cavity obstruction should be treated with high dose of β -blockers as first line therapeutic option. Verapamil or diltiazem could also be considered [1]. Limited experience, mostly from single centers, suggest that mid-ventricular obstruction can be relieved by transaortic or transapical myectomy or by combination of these two approaches [14,15]. Diuretic treatment is contraindicated, because this may lead to LV diameter decrease and consequently to the increase of intraventricular pressure gradient.

Conclusion

AHC is a less common type of HCM. Echocardiography remains the basic diagnostic tool for this form of cardiac disease, nevertheless TTE may underestimate the wall thickness of the apex and apical segments and for this reason diagnosis of AHC may be missed. It can be revealed with the use of 3D echocardiography, however CMR is considered the reference method. ECG recordings taken in ACM patients may mimic acute coronary syndrome due to ST changes.

It should be noted, that adequate hydration is essential and diuretics should be used with caution.

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