Case Report

Coincidence of bicuspid aortic valve presence and hypertrophic cardiomyopathy, and significance of magnetic resonance in its diagnostics

Branislav Obžut*, Peter Blaško, Martin Porzer
Cardiovascular Departement, University Hospital Ostrava, Czech Republic

Abstract
Hypertrophic cardiomyopathy (HCM) is a complex heart disease with a typical pathophysiological characteristic, and with a wide scale of morphologic, functional, and clinical symptoms. The presented case study describes a case of coincidence of a patient with HCM and bicuspid aortic valve, on which we wanted to point out the benefit of the NMR examination and efficiency of alcohol septal ablation with a positive result in a long-term monitoring.

1. Introduction
Hypertrophic cardiomyopathy (HCM) was first described in detail by London forensic pathologist Donald Teare in 1958, who published pathologic findings of young patients with sudden death. The oncoming decades brought a large number of publications describing HCM from many perspectives and labelling this disease with tens of terms—idiopathic hypertrophic subaortic stenosis, muscular subaortic stenosis, or hypertrophic obstructive cardiomyopathy. In the 1970s came globally into use the term “hypertrophic cardiomyopathy”. Historically, the HCM was defined as a heart disease, characteristic with myocardium hypertrophy with consequent absence of any other disease which would initiate the hypertrophy (aortic stenosis, hypertension, amyloidosis, impacts of excessive sports training, etc.). At present, we know that an important proportion of patients suffer concurrently from intraventricular obstruction. Concerning the occurrence of intraventricular obstruction of bloodstream can be in a simplified way said that 1/3 of patients is without the obstruction, 1/3 has latent obstruction which will develop only during stress, and 1/3 of patients has obstruction even in rest conditions. From the genetic point of view, the HCM is in 60% (which is a limitation of this definition) an autosomal dominant disease with incomplete penetration, caused by mutation of genes coding the cardiac sarcomeric proteins. Morphologic image at macroscopic level is characteristic with myocardium hypertrophy, particularly of the left ventricle, which is asymmetric in majority...
of patients. Histopathologic characteristics of HCM include myocyte disarray, disrupted myofibrillar architecture, and defect of intercellular connection with fibrosis. Prevalence of this disease is approximately 0.2% and is thus the most frequent hereditary cardiovascular disease—in the Czech Republic there are approx. 20,000 patients with hypertrophic cardiomyopathy, out of which circa 2000–4000 are carriers of the highest risk of sudden cardiac death [1]. Annual mortality in the non-selected population was reported to be 1% [2]. The most important aspect of the novel interpretation of HCM classification should be in the future particularly the prognostic seriousness of a given HCM form.

The clinical progress of the disease is very diverse. Some patients will remain asymptomatic in the course of their life; with some will develop symptoms of cardiac failure or angina pectoris, while with other patients the sudden cardiac death can be the first display of the disease.

2. Case study

In the presented study is described a case of a 47-year-old male, referred to our workplace with diagnosis of a significant aortic stenosis for selective coronary angiography and other tests, and preparation before cardio-surgical solution of the valve defect. Apart from significant overweight, chronic venous insufficiency, and moderate mixed dyslipidemia, the patient had no important internal anamnesis. The patient has no children and in his family neither this diagnosis was made nor sudden death appeared. The patient used perindopril 10 mg p.o. and metoprolol 50 mg p.o. for about one year. Clinically, the patient showed dominant expressions of cardiac insufficiency in class NYHA III, negated the syncope, stenocardia appeared after a greater stress. ECG examination indicated hypertrophy of left ventricle together with inverted T waves and fixed ST depressions in leads V3-6 and II, III, aVF.

Due to his significant obesity with BMI 45, it was very difficult to examine the patient by transthoracic echocardiogram. In spite of this, there was evident significant hypertrophy of left ventricle with septum thickness 31 mm, the present gradient (gradient at rest)—110 mmHg—was localized in the left ventricle outflow tract (LVOT), not on the aortic valve. As there was suspected presence of bicuspid aortic valve, a trans-oesophageal echocardiography examination was carried out, which confirmed the morphologic finding. Further on was executed NMR examination of heart, confirming the diagnosis of hypertrophic cardiomyopathy with evident SAM (systolic anterior movement) (Fig. 1). From the trans-oesophageal echocardiography, as well as NMR (Fig. 2), was determined the area of the bicuspid aortic valve area (AVA) exceeding 4 cm² (4.5 according to NMR) without serious degenerative changes. The ascending aorta was without signs of dilatation.

After eliminating a significant aortic stenosis and confirming the HCM diagnosis we decided to execute percutaneous transluminal septal myocardial ablation (PTSMA). First the patient underwent an coronary angiography—only unimportant wall...
Fig. 2 – NMRI: Bicuspid aortic valve (with raphe).

Fig. 3 – NMRI: left ventricle hypertrophy after the percutaneous transluminal septal myocardial ablation (PTSMA) (parasternal long axis equivalent projection).
changes were detected. At the beginning of the procedure we applied the echo contrast agent – SonoVue – to display the segment of vascularization of septal branch of the left anterior descending artery. After this verification was applied alcohol using “OTW-balloon” (over the wire). The operation was without complications, the patient was as usual provided with temporary cardiac stimulation by VVI electrode into the right ventricle apex, total amount of the administered alcohol was 1.5 ml, there was a periprocedural gradient decrease (Fig. 3). Already after the completion of the operation, the patient was without conduction disorder, and therefore the temporary cardiac stimulation was cancelled the following day. On the sixth day of hospitalisation the patient was dismissed home.

Apart from the usual echocardiographic checks was executed a control NMR after one month and then after one year (Figs. 4 and 5). After one month, the thickness of myocardium in the place of ablation was 21 mm. After one year of observation, in the place of ablation there was a scar with thickness of myocardium of 16 mm, the subvalvular myocardium thickness was 28 mm and distal myocardium thickness 26 mm (both measurements obtained by NMR imaging).

At present, the patient is in a functional group NYHA II, without stenocardia and by echocardiography with rest gradient 8 mmHg in the left ventricle outflow tract.

3. Discussion

The first percutaneous transluminal septal myocardial ablation (PTSMA) was executed by Ulrich Sigwart in 1995, in the Czech Republic it was Dr. Hlaváček and subsequently Prof. Veselka. This method proves to be safe with the selected group of symptomatic patients with enhancing the quality of life and perhaps also influencing their prognosis.

The coincidence of HCM and bicuspid aortic valve was described already in 1990 by Brown on a set of 4 adult patients [6]. This set did not contain inherent syndromes with which the coincidence of the two units had been known.

In recent years are slowly turning true assumptions pointing out a number of limitations of echocardiography, and on the other hand occurs a number of positives of nuclear magnetic resonance in diagnostics and monitoring of patients with HCM.

Echocardiography remains until now a gold standard in examination of patients’ with HCM heart morphology. A fundamental limitation of echocardiography is the limited ability to examine certain patients, given particularly by obesity or lung diseases with hyperinflation, mainly the lung emphysema. Weak points of the transthoracic echocardiography are the anterolateral segments of the left ventricle, papillary muscles, and right-side heart sections, including diagnostics of right-side intraventricular obstruction. On the other hand, a fundamental limitation of NMR is until now the impossibility to examine patients with implanted cardio stimulator. During examination with utilization of gadolinium, with a normal myocardium occurs its rapid saturation and subsequently a washing-away of contrast. Different situation concerns myocardium with fibrotic foci. In such case occurs an increased saturation by the contrast

![Fig. 4 – NMRI: left ventricle hypertrophy after percutaneous transluminal septal myocardial ablation (PTSMA).](image-url)
medium (gadolinium Gd-DTPA), and by this examination can be detected cicatricial, non-functional foci in myocardium (gadolinium hyperenhancement). Number of such foci and their extent correlate with the IVS thickness, number of hypokinetic segments, lower EFLK, occurrence of positive phenotype already at young age, and occurrence of inconstant ventricular tachycardia. All this indicates the relation of the myocardium fibrosis extent and seriousness of disease including the risk of sudden cardiac death [3].

However, necrosis foci can also be found with a half of patients suffering from hypertrophy of left ventricle as a consequence of aortic stenosis or arterial hypertension [4].

Another group of patients with HCM, which can be identified only with the help of NMR is represented by patients with small, localized hypertrophy focus. Such cases concern patients with apical form of HCM, but also a number of patients with hypertrophy foci in the area of lower or lateral wall – sometimes so called “focal HCM” [5]. It is thus probable that heart NMR will soon become a fundamental examination for patients with HCM or with suspicion on this diagnosis.

**4. Conclusion**

By the described case study of coincidence of a patient with HCM and bicuspid aortic valve, we wanted to point out the benefit of the NMR examination and efficiency of percutaneous transluminal septal myocardial ablation (PTSMA).

In this case, it would be arguable to indicate the patient for a surgical myectomy without aortic valve replacement. Mortality of alcohol septal ablation with a correct execution is 1.5%, and is thus comparable to surgical myectomy [7–9]. At the same time, we are aware that the patient will probably arrive at aortic valve replacement. Nevertheless, in case of wrong indication this would mean a re-operation and thus also a higher periprocedural risk. Heart NMR is a safe and patient-considerate method, which can help to diagnose correctly and determine the correct treatment procedure for the patient.

**References**


