Medical Expertise

"Development of the European Network in Orphan Cardiovascular Diseases"
„Rozszerzenie Europejskiej Sieci Współpracy ds Sierocych Chorób Kardiologicznych"

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CASE SUMMARY

The presented case is a 61-years old White male admitted to the tertiary cardiology centre with the working diagnosis of severe aortic valve stenosis. At presentation, almost a year ago, patients complained about easy fatigue and physical examination has revealed loud systolic murmur radiating to the neck and thoracic scoliosis. Objective assessment of patient's physical capacity, by means of cardiopulmonary exercise test, showed moderately impaired exercise tolerance with peak oxygen uptake of 24 ml/kg/min. On 24-hour tape no serious ventricular arrhythmia was noted but episodes of atrial fibrillation (AF) were recorded. In the initial ecchocardiography (October 2013) hypertrophic cardiomyopathy (HCM) with left ventricular (LV) dynamic obstruction was observed. The LV pressure gradient was very high up to 160 mmHg and caused primarily by systolic anterior motion (SAM) of the elongated anterior and posterior mitral leaflets. Just a month later (November 2013), after institution of beta-blockers (Betaxolol 20 mg), the LV gradient was reduced to 124 mmHg and has been remaining more less stable for almost a year (last echo from August 2014 showed peak LV gradient of 114 mmHg).

DISCUSSION

This is an interesting and educational case of the middle-aged patient with the initial diagnosis of symptomatic aortic valve stenosis admitted with the view of qualification to the aortic valve reaplacement (AVR). However, after detailed echocardiographic assessment it turned to be not an aortic valve disease but HCM with severe LV outflow tract gradient caused by SAM phenomenon. There are few interesting points that need to be taken into account. Firstly, the age of the patient was not typical for first diagnosis of HCM and symptomatic aortic stenosis was absolutely first-line diagnosis. This observation is a perfect example of the importance to be open-minded and be skilled to perform the accurate echocardiography. Secondly, the mechanism of LV obstruction was initially SAM but on the serial echocardiograms the SAM has not been observed any more and yet the LV gradient was still present. This is intriguing observation what actually is causing LV obstruction when SAM effect was diminished. The initial strategy of implementing beta-blockers was absolutely right. However, is it still so after a year of observation and LV gradient being more than 100 mmHg? No wonder, LV cavity has grown bigger and LV wall thickness increased. Although we do not have the recent assessment of patient's functional status, nevertheless, the time has probably come to more

definite mode of treatment. Certainly, an option of changing beta-blockers to non-dihydropiridine calcium channel blockers, such as Verapamil or Dilitiazem, can be contemplated but taking into account only modest effect of beta-blockade, this will have probably similar effect.

EXPERT'S OPINION

The patient's symptoms as well as exercise tolerance should be reassessed. In case the patient is symptomatic, he should be probably referred for the invasive treatment of LV outflow tract obstruction either by means of percutaneous alcohol septal ablation or surgical operation of myomectomy. The final decision regarding the mode of treatment can only be made after meticulous assessment of LV anatomy and mechanisms of LV obstruction. From the already available data, the lack of SAM in the last echocardiograms is in favour of percutaneous treatment. However, when the patient is asymptomatic and his exercise tolerance is not impaired, the decision of invasive treatment is more difficult and not easy. Towards an invasive treatment may be a serial observation of gradual LV cavity dilatation and increase of LV wall thickness. However, these are not verified criteria for intervention and can only serve as a supportive observation. Leaving aside the problem of LV obstruction, one cannot forget about the other serious consequence of HCM, which is premature sudden cardiac death (SCD) caused mostly by ventricular arrhythmias. The new guidelines on HCM, published in September 2014, introduced the new concept of objective assessment of SCD. This is is called the HCM Risk SCD-calculator and gives an exact number of SCD in next 5 years. Based on that figure the further therapeutic decisions, including implantation of cardioverter-defibrillator (ICD) are more objective. The 5-year risk of SCD in this patient is only 2,73%. Therefore, an ICD preventive treatment is not indicated.

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