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 authors presented a case of a 61 years old male, who was admitted to the cardiology department with an initial diagnosis of aortic valve stenosis. He complains of a decrease of exercise capacity. He has arterial hypertension, dyslipidemia and scoliosis. His NT-proBNP level is elevated. ECG reveals signs of left ventricular (LV) hypertrophy. Holter ECG monitoring discloses paroxysmal AFib, episodes of sinus bradycardia and pauses of max duration of 2,7 seconds. Cardiac echo study together with cardiac magnetic resonance reveals LV walls hypertrophy of max 25mm, normal size chambers, LV outflow tract obstruction (LVOTO) due to mitral anterior leaflet systolic anterior motion (SAM) causing max LVOT gradient of around 120 - 160mmHg, moderate AV regurgitation. Cardiopulmonary exercise test shows mildly-to-moderately reduced exercise capacity with VO2max of 24ml/kg.min. The patient is administered VKA, sotalol 20mg/d, amlodipine 10mg/d, ACEi 10mg/d and statin. The authors are uncertain about the most appropriate further management.

DISCUSSION

HCM is defined by a wall thickness ≥15 mm in one or more LV myocardial segments, that is not explained clearly by loading conditions. The prevalence of unexplained increase in LV thickness ranges between 0.02–0.23%. 40-60% of HCM cases is caused by sarcomeric protein mutation, 5-10% by other genetic and non-genetic causes including; metabolic, mitochondrial, neuromuscular, infiltrative or endocrine disorders, and in up to 30% of cases the origin remains unknown. Clinical presentation depends on the underlining cause. Symptoms include chest pain, chronic heart failure, syncope and heart palpitations. Management of HCM patients should include i.a.

assessment of symptoms, laboratory testing for detection of extra-cardiac conditions and the severity of cardiac involvement (NT-proBNP and high sensitivity cardiac troponin T), 12-lead ECG for detection of rhythm abnormalities, imaging (echo, CMR) for defining LV systolic and diastolic function, presence of left ventricular outflow tract obstruction (LVOTO), morphology and function of other heart chambers and valves, presence of myocardial fibrosis (CMR), cardiopulmonary exercise testing for assessment of exercise capacity, family history to guide genetic testing and the assessment of risk of sudden cardiac death.

Up to 60% of HCM patients have LVOTO. One third of them have resting systolic anterior motion (SAM) of the mitral valve leaflets, while another third have latent obstruction only during manoeuvres that change loading conditions and LV contractility. LVOTO is defined as Doppler LVOT gradient of ≥30 mm Hg, with the threshold for invasive treatment of more ≥50 mm Hg. In symptomatic patients with LVOTO, the aim is to improve symptoms by using drugs, surgery, alcohol ablation or pacing.

EXPERT'S OPINION

This is an elderly patient with HOCM with max LVOT gradient of >100mmHg. He additionally suffers from not optimally controlled arterial hypertension. He seems to be mildly symptomatic. According to the ESC guidelines on management of patients with HCM (2014) Septal reduction therapy to is recommended in patients with a resting LVOT gradient of > 50 mm Hg, who are in NYHA functional Class III–IV, despite maximum tolerated medical therapy. The mode of intervention depends on the experience of the centre. This patient seems not to be in NYHA class III or more, therefore septal reduction therapy is not recommended in this patient at this point. Pharmacological treatment should include non-vasodilating betablocker (eg. sotalol) to improve symptoms and to control arrhythmia. Vasodilationg agents like amlodipine and ACEi should are not recommended in patients with HCM and should be used with caution to control hypertension in this patient. Cardiac pacing should be consider in this patient to prevent bradycardia and to enable up-titration of betablocker dosing. ICD implantation is not recommended in this patient based on ESC HCM SCD risk formula – calculated risk of SCD at 5 years is 3,2.

Coronary angiography should be considered in this patient to assess the coronary arteries anatomy together with septal branches. Broadening of the search for identifiable causes of HCM like Fabry diseases or amyloidosis is advised. Family screening should be carried out. The patient should remain in follow-up with assessment of symptoms, ecg, echo, exercise test and laboratory testing every 6-12 months.

1. Conservative treatment should be recommended in this patient at this point.

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2. Septal reduction therapy is not recommended at this point
3. Non-vasodilating betablockers should be used.
4. Management of arterial hypertension with amlodipine and ACEi should be conducted with caution
5. Cardiac pacing should be consider in this patient to prevent bradycardia and to enable up-titration of betablocker dosing.
6. ICD implantation is not recommended in this patient.
7. Coronary angiography is advised to assess coronary arteries anatomy.
8. Assessment of identifiably causes of HCM should be performed.
9. Family screening is advised.
10 6-12 months follow-up with assessment of symptoms, ecg, echo, exercise test and laboratory testing is advised.

REFERENCES

Elliott PM, Anastasakis A, Borger MA et al. 2014 ESC Guidelines on diagnosis and management of hypertrophic cardiomyopathy: The Task Force for the Diagnosis and Management of Hypertrophic Cardiomyopathy of the European Society of Cardiology (ESC). Eur Heart J. 2014;35:2733-79.

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