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Case report

Hypertrophic cardiomyopathy: from accidental diagnosis to sudden cardiac death prevention

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Abstract Hypertrophic cardiomyopathy is defined by the presence of increased thickness of the left ventricular wall, which cannot be explained only by abnormal loading conditions. This definition applies to children and adults and makes no „a priori „assumptions about etiology or myocardial pathology. Cellular architecture disorders, interstitial fibrosis, microvascular infarctions, play a role in the emergence of electrical instability, malignant ventricular arrhythmias that cause Sudden Cardiac Death in patients with HCM. Although most cases have a benign prognosis, identifying patients at a risk for Sudden Cardiac Death, requiring prophylactic therapy with ICD ,is crucial and prioritized. With increasing awareness of the disease, lower-risk patients are now more frequently diagnosed, and more recent studies show that the annual Sudden Cardiac Death rate 0.5-1%, was not negligible,unfortunately among young and asymptomatic individuals. This paper presents a case of HCM, accidentally detected in a young adult, completed with ICD implantation in primary Sudden Cardiac Death prevention .

Kew words hypertrophic cardiomyopathy, sudden cardiac death

Introduction

Cardiomyopathies have been defined as structural and functional abnormalities of the ventricular myocardium that cannot be explained by disease due to limited flow in the coronary arteries or pathological loading conditions[1]. Historically, this group of disorders has been divided into primary diseases, in which the heart is the only nvolved organ , and secondary forms where cardiomyopathy is the manifestation of a systemic disorder. The guidelines of the European Society of Cardiology (ESC) define cardiomyopathies by specific morphological and functional criteria, and then group them into family / genetic and non-familial / non-genetic subtypes, regardless of the presence of some other heart diseases.

Hypertrophic cardiomyopathy (HCM) is defined by the presence of increased left ventricular (LV) wall thickness that cannot be explained by abnormal loading conditions alone. This definition applies to children and adults and makes no " a priori " assumptions about the etiology or myocardial pathology.[1]

Cellular architecture disorder, interstitial fibrosis, microvascular infarctions play a role in the onset of electrical instability, ie malignant ventricular arrhythmias that cause sudden cardiac death / SCD / in patients with HCM.[2]

In up to 60% of adolescents and adults with HCM, the disease is an autosomal dominant genetic trait caused by mutations in the heart sarcomer protein genes.[3,4,5,6] Five to 10 percent of adults with HCM are caused by other genetic disorders, including hereditary metabolic and neuromuscular diseases and chromosome abnormalities.

In adults, HCM is defined by a wall thickness of ≥15mm in one or more LV segments - measured by any imaging technique (echocardiography, cardiac magnetic resonance imaging (CMR) or computed tomography (CT)), which cannot be explained exclusively loading conditions. Genetic and non-genetic disorders can be represented by a lesser degree of wall thickening (13-14mm) - in these cases, the diagnosis of HCM requires evaluation of other elements, including family history, non-cardiac symptoms and signs, pathologic electrocardiogram (ECG), laboratory tests, and multiple cardiac modalities of imaging.

Age are one of the most important factors to consider a possible cause of HCM. For example, hereditary metabolic disorders and congenital dysmorphic syndromes are much more common in infants than in older children or adults. Creating a family history of three to four generations helps to confirm the genetic origin of the disease and identify other family members who are at risk of developing the disease. Specificities to be noted in family history include: SCD, unexplained cardiac arrest, heart transplantation, implanted pacemaker and defibrillator and evidence of systemic disease (young age, skeletal muscle weakness, kidney dysfunction, diabetes, deafness, etc.). crucial for the diagnosis and monitoring of HCM. In most patients, hypertrophy primarily involves the interventricular septum in the basal segments of the LV, but often extends to the lateral wall, posterior septum, and apex.[7]

CMR should be considered in patients with HCM in their baseline assessment if local resources and expertise allow. In patients with good echocardiographic imaging, CMR provides similar information on ventricular function and morphologies and[8,9], but is useful in establishing a

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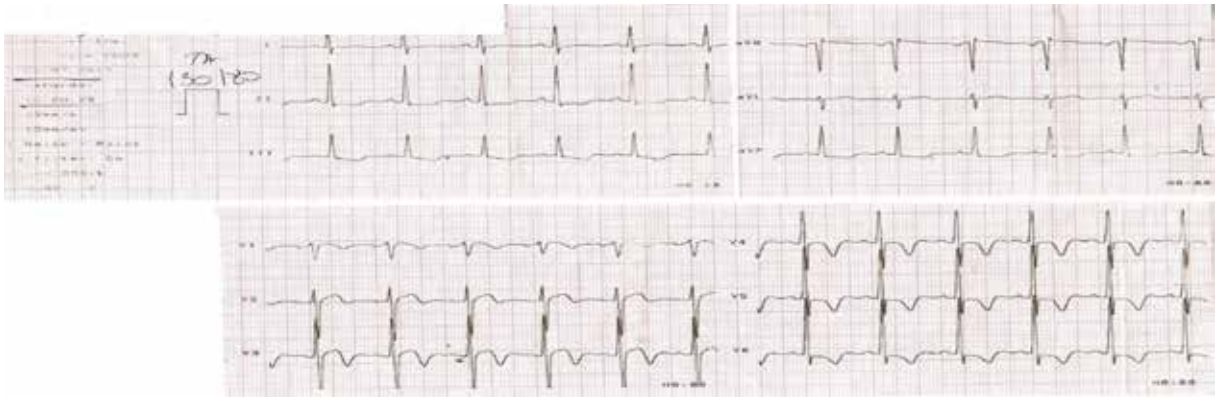


Figure 1. ECG on admission

diagnosis of HCM in patients with poor acoustic window or when some LV regions are poorly visible - such as the anterolateral wall , top of LV and right ventricle.[10,11]
Most people with HCM are asymptomatic and have a normal lifespan, but some develop symptoms, often many years after the onset of ECGs or echocardiographic signs of HCM. SCC, heart failure, and thromboembolism are major causes of death[12] The most commonly reported fatal arrhythmic event is spontaneous ventricular fibrillation (VF), but asystole, AV block, and electromechanical dissociation have also been reported. [13,14,15,16,17] The disease is mostly asymptomatic and is discovered by accident.

Case presentation

Patient aged 31, hospitalized in our institution in July 2019 after seeing ECG changes in the type of negative T waves, predominantly precordial with frequent palpitations, rapid fatigue. Symptoms were presented since November 2018 when his brother, a born aunt, an athlete, died in sleep at the age of 29. Autopsy finding: "HCM and most likely arrhythmic event as cause of death. Heart muscle thickened to about 23 mm".
The patient has no risk factor for ischemic heart disease, denies any chronic diseases and regular medication. ECHOcardiographically revealed concentric hypertrophy of the left ventricular wall, with 17 mm thick IVS, pseudoSAM movement of the anterior mitral leaf, with no significant gradient in the left ventricular outflow tract / LVOT /. and smaller, mild MR with preserved global left ventricula systolic function. EF 65%.

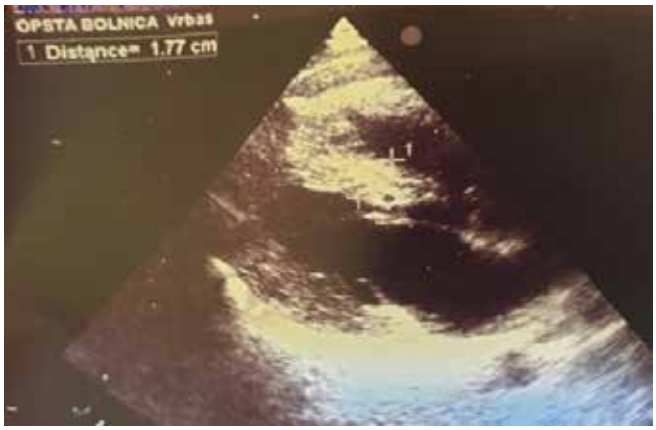
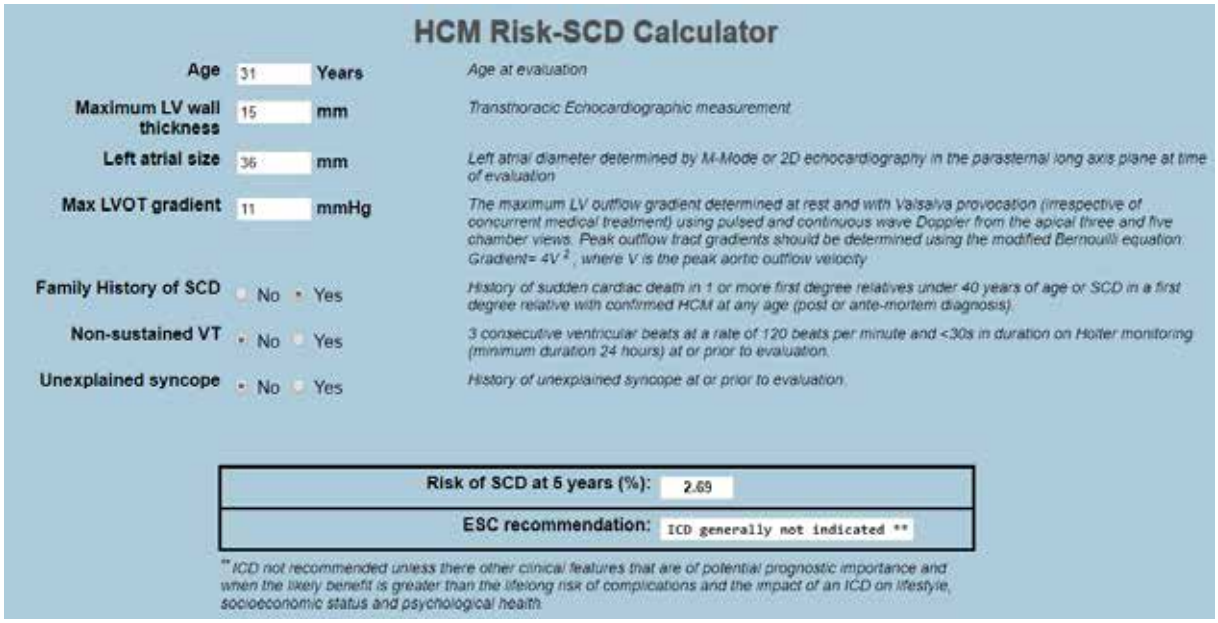


Figure 2. ECHOcardiography PLAX position in maximal diastole

Clinical diagnosis of HCM in the first degree of relatives of patients with clear disease (HCM ≥15 mm) is based on the presence of an unexplained increase in LV wall thickness ≥13 mm in one or more LV myocardial segments as measured by any cardiac imaging technique. Holter monitoring doesn t show heart rhythm disorders. The patient's medical records were presented to the Cardiology council in a tertiary institution,which indicating an expert Echocardiographic examination and MSCT coronarography. Cardiopulmonary compensated, rhythmically stable, with low dose / 2.5 mg / of cardioselective Beta blocker,patient was released for further home treatment from our hospital. Expert heart ultrasound done for better visualization of the endocardium, using transpulmonary contrast with a registered IVS thickness of 15 mm. MSCT coronarography with neat finding.CMR scan detected moderately pronounced myocardial hypertrophy, without focal zones of pathological signal. Stress echocardiography (SEHO) finding with neat finding. CFR for RCA preserved 2.87 CFR flow reserve for LAD preserved 2.81

Should we suggest this patient to incorporate ICD in primary SCD prevention?
Patients who have previously had abortion SCC and malignant ventricular arrhythmias are at greater risk for further arrhythmic events, and in such patients implantation of ICD is not disputed. However, the choice of patients to receive ICD for primary prevention of ISS is more difficult.
There are very few randomized, controlled, clinical studies in patients with HCM, and therefore most current ESC recommendations are based on the findings of observational retrospective cohort studies and consensus of expert opinions. In one study conducted by Spirito et al., which included 668 patients with HCM without conventional risk factors, assymptomatic or with mild symptoms, the risk of SCD was not negligible - a rate of 0.6% per year. (**18**)
This finding therefore underscores the importance of stratifying the risk of SCC in patients with proven HLV. Since 2014, the ESC has recommended the use of a new quantitative risk score / HCM Risk-SCD /, composed of seven disease-related characteristics, to predict ISS over a five year period. Based on the results, patients were stratified into three subgroups for recommendation to install ICDs in primary prevention of SCC. Scor <4% indi-



cated as low and in which the implantation of ICD is not indicated, Scor 4-6% indicated as median , may consider implantation of ICD ,while group with a score of ≥6% indicates implantation of ICD.

HCM Risk-SCD Score 2.69 classified our patient to a Low risk group for SCD, and the installation of ICD should not be considered, but continued subjective status monitoring with more frequent Holter monitoring. However, it was decided to seek the opinion of an eminent arrhythmologist , after the searches had been carried out. Opinion was stated in one sentence: **„ICD, without delay".**

Conclusion

Although most cases of HCM have a benign prognosis, identifying patients at risk for SCD requiring prophylactic therapy with ICD, is crucial and prioritized. With increasing awareness of the disease, lower-risk patients are now more frequently diagnosed, and more recent studies show that the annual rate of SCD is 0.5-1% , unfortunately among young and asymptomatic individuals.

There are very few randomized, controlled, clinical studies in patients with HCM, and therefore most current ESC recommendations are based on the findings of observational retrospective cohort studies and the consensus of expert opinions.

In this case, the expert opinion of an Eminent arrhythmologist was sought and the patient was implanted with an ICD device in primary prevention of SCD . The patient is enrolled in one of the clinical studies ,that will allow additional genetic testing through participation.

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Sažetak

Hipertrofična kardiomiopatija: od slučajne dijagnoze do prevencije iznenadne srčane smrti

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Hipertrofična kardiomiopatija je definisana prisutnošću povećane debljine zida leve komore, što se ne može objasniti samo nenormalnim uslovima opterećenja. Ova se definicija odnosi na decu i odrasle i ne daje „a priori" pretpostavke o etiologiji ili miokardnoj patologiji Poremećaji ćelijske arhitekture, intersticijalna fibroza, mikrovaskularni infarkti igraju ulogu u nastanku električne nestabilnosti tj malignih ventrikularnih aritmija koje uzrokuju iznenadnu srčanu smrt u bolesnika sa Hipertrofičnom kardiomiopatijom. Iako je u većini slučajeva prognoza benigna, prepoznavanje bolesnika s rizikom Iznenadne Srčane Smrti ,koji zahtevaju profilaktičku terapiju ICD-om, presudno je i prioritetno. S povećanjem svesti o bolesti, pacijenti sa nižim rizikom sada se dijagnostikuju više, a novija istraživanja pokazuju da godišnja stopa iznenadne srčane smrti iznosi 0,5-1% što nije zanemarljivo, nažalost među mladim i asimptomatskim pojedincima. U ovom radu prikazan je slučaj HCM-a, slučajno otkriven u mlađe odrasle osobe sa pozitivnom porodičnom anamnezom,završen implantacijom ICD-a u primarnoj prevenciji iznenadne srčane smrti

Ključne reči: hipertrofična kardiomiopatija, iznenadna srčana smrt