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Myocardial Perfusion Abnormalities in Patients with Hereditary Hypertrophic Cardiomyopathy: A Study of Three Cases and Review of the Literature

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Abstract

Introduction: Hypertrophic cardiomyopathy (HCM) belongs to the very heterogeneous group of cardiomyopathies. This study aimed to study myocardial perfusion abnormalities on scintigraphy and assess the risk of sudden death from ventricular arrhythmia in hereditary sarcomeric HCM. **Patients and Methods:** This is a retrospective and prospective descriptive study over 18 months (January 01, 2021, to July 31, 2022) on the records of patients over 18 with the diagnosis of hypertrophic sarcomeric cardiomyopathy with or without ventricular rhythm disorders and who have undergone myocardial scintigraphy. **Results:** Three patients were included. The average age of our patients was 66 years old. Dyspnea is the primary symptomatology found in our patients. One patient presented with syncope and unsustained ventricular tachycardia. His risk score for sudden death from ESC at five years is estimated at 6.45%, and the patient received an ICD in primary prevention. The average sudden death risk score of our patients was 3.78%. The mean LV wall thickness was 20 mm. The mean maximum left intraventricular gradient was 39 mmHg. Myocardial fibrosis was present in all our patients. Myocardial scintigraphy was normal in all cases. **Conclusion:** Hypertrophic cardiomyopathy is a very heterogeneous group of cardiomyopathies. The rhythmic risk is multifactorial and constitutes a significant prognostic factor.

Keywords

Hypertrophic Cardiomyopathy, Myocardial Scintigraphy, Sudden Death, Ventricular Arrhythmia

1. Introduction

Hypertrophic cardiomyopathy (HCM) belongs to the very heterogeneous group of cardiomyopathies. These are diseases associated with damage to the heart muscle with no apparent cause [1] [2]. This entity was first described in the late 1950s [1] [3]. It is characterized by hypertrophy, *i.e.* excessive thickening, of the heart wall [1] [4]. It predominantly affects the left ventricle [1] [4]. Prevalence is 0.2% in adults and 0.3 - 0.5 per 100,000 children in the general population [1] [5] [6].

The disease is usually genetic in origin, most often caused by mutations in sarcomere genes (in 40% - 60% of cases), with transmission usually autosomal dominant [1] [2].

Non-sarcomeric causes have been identified, occurring in around 25% of children and 10% of adults, prompting systematic referral to an expert center at the time of diagnosis [1] [7]. It is associated with the risk of sudden death. In the United States, HCM is the leading cause of sudden death in young athletes under 40, as reported by MARON and colleagues [8] [9]. Despite the enormous progress made in this field, risk stratification of sudden death in patients with HCM remains a challenge [10] [11].

Although ventricular arrhythmias are responsible for most of these events, the mechanisms responsible for cardiac arrest in HCM are not understood. Abnormalities in myocardial perfusion may create an arrhythmogenic substrate in patients with HCM [10].

The aim of this study was to investigate myocardial perfusion abnormalities on scintigraphy and assess the risk of sudden death due to ventricular rhythm disturbance in hereditary sarcomeric HCM.

2. Patients and Methods

1) Framework and period of study

The cardiology department of the Center Hospitalier Intercommunal Alençon-Mamers (CHICAM) served as a framework for the realization of this study. The study occurred from January 01, 2021, to July 31, 2022.

2) Type of study

This is a retrospective descriptive study on the records of patients diagnosed with sarcomeric hypertrophic cardiomyopathy with or without ventricular arrhythmias.

3) Study population

Patients admitted to the department with confirmed hypertrophic cardi-

omyopathy.

Inclusion criteria

All patients over 18 years of age were diagnosed with hypertrophic sarcomeric cardiomyopathy and underwent myocardial scintigraphy.

Non-inclusion criteria

Patients with HCM who did not have myocardial scintigraphy or with an uninterpretable examination.

4) Variables studied

The variables studied were: socio-demographic (age, gender); clinical (right or left heart failure, congestive heart failure, chest pain, syncope or faintness, dyspnoea); electrocardiographic (search for heart rhythm and conduction disorder); rhythmic holder (search for paroxysmal rhythm disorder); stress test; echocardiographic (for diagnosis, search for hypertrophy, evaluation of my LVEF, search for SAM, intraLV gradient); cardiac MRI for diagnostic confirmation, research of my fibrosis; myocardial scintigraphy for perfusion abnormalities.

Our results are presented in text form. We have expressed qualitative variables as percentages and quantitative variables as mean plus or minus standard deviation.

3. Results

We included three patients, two men and one woman. The average age of our patients was 66 years old, with extremes of 52 and 79 years old. All our patients (100% of cases) had dyspnea. None of the patients had shown signs of congestive heart failure. One of our patients presented with typical syncope. All our patients (100% of cases) had electrical left ventricular hypertrophy with repolarization disorders. The average Sokolow index was 50 mm, with extremes of 45 mm and 55 mm—one patient presented with bursts of monomorphic ventricular tachycardia at follow-up. The stress test was without abnormality in our patients.

On the echocardiographic level, the average left ventricular hypertrophy was 20 mm with extremes of 19 mm and 21 mm. All our patients had a preserved left ventricular ejection fraction with an average of 59%. We noted an alteration in the global longitudinal strain (GLS); the average GLS was −14%, with extremes of −12% and −17%. The average intraventricular gradient was 39 mmHg with extremes of 11 mmHg and 60 mmHg.

SAM had been found in 100% of cases with minimal to moderate mitral leakage and a saber blade appearance (**Figure 1**). Only one patient had an insertion anomaly of the lateral pillar of the anterior mitral valve.

The left atrium was dilated in all our patients with an average anteroposterior diameter of 48 mm with extremes of 45 mm and 53 mm. Cardiac MRI found intramyocardial fibrosis in two patients (66%). The average thickness of the SIV was 19 mm with extremes of 17 m and 23 mm. One patient had predominantly apical LVH. All of our patients had normal myocardial scintigraphy (**Figure 2**).

Evolutionarily, one of our patients presented a monomorphic unsustained

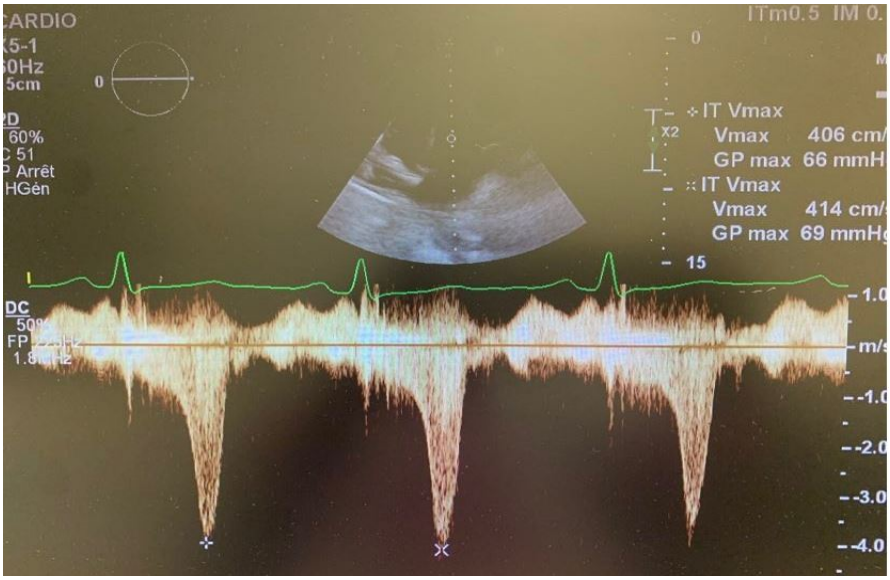

Figure 1. Echocardiography image in continuous Doppler mode with saber blade appearance.

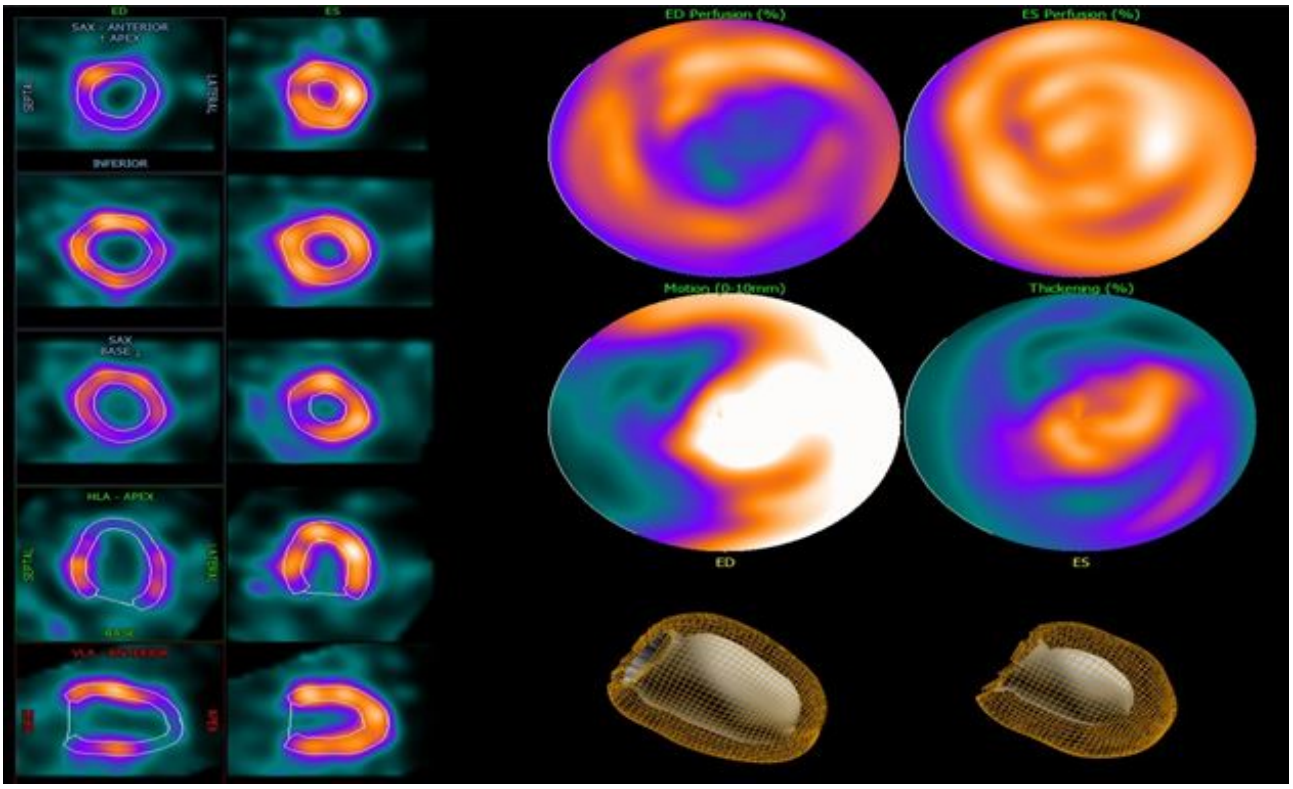

Figure 2. Myocardial scintigraphy image without significant perfusion abnormalities.

ventricular tachycardia on rhythm holter and an episode of syncope during follow-up. The risk score for sudden death from ESC (HCM risk score) was 6.45%. Our patient benefited from an implantable cardioverter defibrillator (ICD) in primary prevention without subsequent rhythmic events during follow-up. Our two other patients had a low-risk score for sudden death from ESC (HCM risk

score) of 2.74% and 2.17%, respectively, allowing them to avoid the DAI. Our patients' average HCM risk score was 3.78%, with extremes of 2.17% and 6.45%. No patient presented with sudden death during follow-up. All our patients were on beta-blockers with Propranolol in two patients and Sotalol in one patient.

4. Discussion

The size of our sample, the geographical accessibility of myocardial scintigraphy, the retrospective nature and the short duration of the prospective part were our main limitations and constraints in this study. The average age of our patients was 66 years, with extremes of 52 years and 79 years, with a male predominance of 66%. This result is close to that of Sarr in Dakar [12], who found an average age of 53 with extremes of 27 and 79 years. As for Maron [13], he had found an average age of 34 years. Clinically, not all of our patients had shown signs of congestive heart failure. Our results corroborate those of the literature, particularly that of Charron *et al.* [3], who found left ventricular dysfunction and congestive heart failure in 10% of cases over ten years-however, one in three patients presented with typical syncope. Electrical LVH was present in all our patients and repolarization disorders. This observation is classic in this pathology.

On the other hand, Sarr [14] found LVH in 68.7% of cases and repolarization disorders in 56.2% of cases despite the high frequency of repolarization disorders in black subjects, particularly athletes, found in the literature. One patient presented with unsustained ventricular tachycardia on rhythm holter. This ventricular arrhythmia is classic in this condition and more often not sustained [3] [14]. This is why it is essential to perform electrical monitoring to assess the rhythmic risk in these patients objectively. All our patients had a standard test with no arrhythmia and low blood pressure. This result is almost identical to that of Sarr [12], who mentioned a blood pressure mismatch in a 27-year-old patient out of a sample of 16 patients. The maximum mean left ventricular wall thickness of our patients was 20 mm. This result is identical to those of Sarr [14] and Maron [15], who found 20.9 mm and 21 - 23 mm, respectively. The left ventricular ejection fraction was preserved with an average of 59%. As described in the literature, the left ventricular ejection fraction (LVEF) remains most often preserved, which is the case in our study.

All our patients had an alteration of the GLS. This alteration of the GLS is constant in the CMH, which encourages the systematic performance of a GLS before any LVH, even in hypertensive patients [2]. Most often, there is a conservation of regional strain in patients with post-hypertensive LVH. All our patients had a maximum left intraventricular gradient; the average was 39 mmHg with extremes of 11 mmHg and 60 mmHg at rest. Anterior systolic movement of the mitral valve (AMS) was found in 100% of cases with minimal to moderate mitral leakage; on the other hand, Sarr [14] found a percentage of 18.8% lower than that of our series. This encourages us to keep going with the diagnosis of HCM in its absence.

Charron [3] noted the presence of SAM, the importance of which is correlated with a systolic pressure gradient. On Doppler, he found a characteristic ejection flow with progressive acceleration and peak end-systolic velocity ("sabre blade" appearance) and mitral insufficiency is sometimes noted. Only one patient had an insertion anomaly of the lateral pillar of the anterior mitral valve. This inconstancy of mitral pillar insertion anomaly should not systematically refute the diagnosis of HCM in cases of LVH without apparent cause.

Dilatation of the left atrium was present in all our patients (100% of cases), and the average anteroposterior diameter was 48 mm with extremes of 45 mm and 53 mm. Our results can be superimposed on those of Sarr [14], who found in his study a dilation of the left atrium in 87.5% of cases. The cardiac MRI allowed us to find two cases of intramyocardial fibrosis. This myocardial fibrosis is correlated with the rhythmic risk in these patients, unfortunately not considered in the risk score for sudden death from ESC at five years.

Myocardial scintigraphy did not show any ischemia in our patients.

This result could be partly explained by the low frequency of severe ventricular arrhythmias in our patients with a low average risk score of sudden death from ESC of 3.78% against prophylactic implantation of the ICD. Nevertheless, a prospective study with many patients will be needed to detect myocardial ischemia, probably responsible for the malignant ventricular disorder during exercise and whose correction was made by prescribing an anti-anginal vasodilator treatment-only one patient presented with unsustained ventricular tachycardia and an episode of syncope. The 5-year sudden death risk score for ESC was evaluated at 6.45%, and the patient received an ICD without recurrence during rhythm monitoring.

5. Conclusion

Hypertrophic cardiomyopathy is a very heterogeneous group of cardiomyopathies. The rhythmic risk is multifactorial and constitutes a significant prognostic factor. We found no myocardial ischemia on scintigraphy to assess the rhythmic risk. Nevertheless, it will take a prospective study with many patients to authenticate myocardial ischemia as a risk factor for malignant ventricular rhythm disorder and, therefore, the indication for defibrillator implantation in primary prophylaxis.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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