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# HYPERTROPHIC CARDIOMYOPATHY WITH OBSTRUCTION OF THE LEFT VENTRICLE

**Bryam Esteban Coello García[1], Andrea Estefanía Cañar Mendes[2], Sebastián David Orellana Méndez[3], Diego Javier Cañar Calderón[4], Karla Viviana Izurieta Duche[5], Galo Fernando Tulcanaza Ochoa[6], Darwin Arturo Pullas Medina[7]**

*[1]General Practitioner in independent practice, Faculty of Medical Sciences, Universidad de Cuenca. Azuay - Ecuador. ORCID: https://orcid.org/0000-0003-2497-0274*

*[2]General Practitioner in Primary Health Care - Ecuador. ORCID: https://orcid.org/0000-0003-3205-8074*

*[3]General Practitioner in SOLCA - Cancer Institute. Cuenca - Ecuador. ORCID: https://orcid.org/0000-0003-0774-1250*

*[4]General Practitioner in independent practice, Faculty of Medical Sciences, Universidad de Cuenca. Azuay - Ecuador. ORCID: https://orcid.org/0000-0002-1496-3663*

*[5]General Practitioner in Primary Health Care - Chile. ORCID: https://orcid.org/0000-0002-3909-0088*

*[6]General Practitioner in "Hospital Misereor" of Gualaquiza. Morona Santiago - Ecuador. ORCID: https://orcid.org/0000-0002-6254-8624*

*[7]General Practitioner in Primary Health Care - Ecuador. ORCID: https://orcid.org/0000-0002-9461-8535*

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## SUMMARY

***It is a septal hypertrophy, asymmetric type, causing left ventricular outflow obstruction due to thickening of the left ventricular wall; this pathology has a worldwide prevalence of 0.02-0.23%; and it is the most important cause of sudden death in young people. It is characterised by myocardial hypertrophy leading to diastolic dysfunction; epidemiologically, it occurs equally in men and women, although women tend to be more symptomatic, more disabled and present at a younger age.***

***The patient may be asymptomatic or may present with dyspnoea, precordial pain and syncope. For diagnosis, chest X-ray, electrocardiogram and echocardiography may be used; MRI and cardiac catheterisation only in cases to identify the aetiology and severity of the disease. For treatment, background medication such as beta-blockers may be used, or alcohol ablation techniques may be performed. However, in some cases, pacemakers or implantable defibrillators may be needed.***

***The aim of this literature review is to provide information on this pathology, which, despite not being so common in our environment and being more of a speciality of Cardiology, there is the possibility of presentation in young people, and that, presenting cardiac manifestations, can lead us to consider this pathology as a differential diagnosis.***

**KEYWORDS**: ***Cardiomyopathy, septal hypertrophy, sudden death, diastolic dysfunction***

## INTRODUCTION

Around 35-60% of patients with this pathology have a mutation in the genes encoding the sarcomeres (1); mostly related to hereditary family transmission; non-genetic causes are observed in 25% of patients, possibly related to errors in metabolism, such as Pompe disease, malformations, and neuromuscular disorders, such as Friedrich's Ataxia (2) (3). In adults, the prevalence of non-genetic causes is low, but if it were to occur, it would be related to Denon's disease, Fabry's disease, left ventricular hypertrophy associated with Wolff-Parkinson-White disease. The prevalence of this disease is estimated to be about 1 in 500 in the general population. In children it is unknown but an incidence of 0.3 - 0.5 per 100,000 is reported (4) (5). Transmission of this disease is genetic, 50% in each subsequent generation, due to mutation

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of at least 1 of 11 genes; in case of no family history, transmission of the disease is generated by a new mutation (6).

## PATHOPHYSIOLOGY

When this cardiomyopathy is accompanied by altered myofibrillar structure and fibrosis, it causes diastolic dysfunction, haemodynamic alteration, including asymmetric ventricular relaxation, decreased cavity filling and abnormal intracellular calcium consumption. Because the coronary vessels fill during diastole, and if there is outflow tract obstruction and ventricular stiffness, there is an increased risk of myocardial ischaemia, leading to ventricular arrhythmias and sudden death. It mostly affects the ventricular septum.

This cardiomyopathy can be classified as obstructive or non-obstructive. This cardiomyopathy can be classified as obstructive or non-obstructive. Depending on the degree of obstruction and the clinical presentation, the degree of hypertrophy depends (7)(8).

Dynamic outflow obstruction is generated by the anterior systolic movement of the anterior leaflet of the mitral valve, as the leaflets impact on the hypertrophied basal septum, causing a pressure gradient that pulls the anterior leaflet, causing an obstruction of the outflow tract, generating a turbulent flow at the time of contraction, decreasing the flow and causing mitral regurgitation. In the case of non-obstructive type, it generally has a good prognosis, with symptoms originating from diastolic dysfunction, except if there is thinning of the wall, dilatation of the left cavity and systolic dysfunction, together with secondary pulmonary hypertension, in which case it would have a poor prognosis. In 25% of patients, obstruction occurs at rest, but in 70% of patients with provocative manoeuvres, such as exercise. In more advanced stages, this may occur at rest. More commonly, it occurs with provocative manoeuvres such as exercise due to increased myocardial demand (9).

## CLINICAL MANIFESTATIONS

The clinical picture can manifest itself in a variable way; it can even be asymptomatic and can be identified accidentally. However, it may manifest with Atrial Fibrillation in 1 out of 5 patients, thus presenting a risk of arrhythmias and subsequent decompensation; therefore, treatment is based pharmacologically with anti-coagulants to prevent the formation of emboli (10).

When there is significant haemodynamic obstruction, dyspnoea may occur, due to elevated filling pressure caused by diastolic dysfunction, outflow tract obstruction, mitral regurgitation and myocardial ischaemia, resulting in fatigue and syncope due to reduced blood supply, eventually progressing to heart failure (11). Ultimately, when dyspnoea, precordial pain, presyncope and syncope occur gradually, they are associated with poor prognosis in the future (12).

## DIAGNOSIS

- Chest X-ray: identifies left ventricular hypertrophy.
- Electrocardiogram: shows left ventricular hypertrophy. It may also show arrhythmias, although it can often be normal.
- Echocardiography: with diagnostic accuracy of 80% for Obstructive Hypertrophic Cardiomyopathy. It can visualise the size and make measurements of the septum and left ventricular wall. It can demonstrate flow velocity along the left ventricular outflow tract.
- Colour Doppler may show mitral valve regurgitation.
- Cardiac magnetic resonance imaging: Gold standard for the diagnosis of left ventricular wall properties
- In patients with suspected Obstructive Hypertrophic Cardiomyopathy with a normal or inconclusive echocardiogram.
- Cardiac catheterisation: only to accurately determine the degree of outflow tract obstruction, the anatomy of the left ventricle, the degree of mitral valve insufficiency and the patency of the coronary arteries (13)(14).

## TREATMENT

Beta-blocker therapy improves myocardial oxygen delivery, reducing ischaemia and prolonging diastolic blood filling and slowing the heart rate. If beta-blockers are ineffective or not tolerated, calcium channel blockers may provide symptomatic relief. In patients with refractory symptoms, septal reduction therapy, either with myomectomy or alcohol septal ablation, represents the second line of therapy for patients with persistent left ventricular outflow tract obstruction (greater than or equal to 50 mmHg).

The indication for alcohol-based septal ablation is: advanced age, presence of comorbidities that would increase the risk of cardiac surgery, history of previous cardiac surgery, failed myomectomy or previous stroke. Mortality 30 days after the procedure is very low (0.6%), and annually, the probability of death is the same as in the general population (15)(16)(17).

However, when myomectomy is performed and its outcome is inadequate, the need for a pacemaker is imminent post-operatively; further demonstrated in procedures performed at the Mayo Clinic in the United States, where it was shown that, at the end of the ablation procedure, there was a high prevalence of early mortality and the presence of perioperative morbidity, including the use of a pacemaker secondary to heart block, despite successful improvement of the outflow tract gradient. In the case of transthoracic septal myomectomy in young adults and adolescents, the gradient improvement was 89 mmHg, although they had systolic anterior motion with mitral regurgitation, so a decision was made between implantable defibrillator placement in 17% or permanent pacemaker in 11.7%. In addition, performing this procedure increases the prevalence of iatrogenic morbidity, thus requiring a pacemaker (18) (19).

In recent years, there has been the development of new drugs and innovative therapies, which are an important aid to the pathophysiological treatment of the disease, including:

- Diltiazem, which inhibits the entry of calcineurin into the sarcomere
- Ranolazine, which inhibits the flow into the sarcomere and increases the outflow of calcium via sodium-calcium exchange.

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- MYK-461: small molecule inhibitor of myosin ATPase, which prevents the cross-bridge cycle produced by myosin.
- NAC: N-acetylcysteine, which reduces glutathionylated myofilament levels with reversal of increased myofilament calcium sensitivity, diastolic dysfunction, myocyte hypertrophy and fibrosis.
- Perhexiline: a myocardial metabolic modulator that changes cellular metabolism to promote more efficient carbohydrate metabolism by inhibiting mitochondrial free fatty acid uptake and utilization, thereby improving myocardial efficiency.
- Angiotensin receptor blockers: decreases the tropic factors of production of the renin-angiotensin aldosterone system pathway (20).

## CONCLUSIONS

Hypertrophic cardiomyopathy is a pathology based on septal hypertrophy, which generates an obstruction of the outflow of the left ventricle, which generates a decrease in ventricular filling, thus generating symptoms; there is a non-obstructive form, which has a good prognosis unless risk factors are present. It is an important cause of death in women and young people. The aetiology is usually hereditary in 50% and mutations when it is not hereditary. In addition, this pathology is associated with diseases such as Fabry's disease, Denon's disease, and Wolf Parkinson White. It usually manifests with dyspnoea, fatigue, pre-syncope, syncope and atrial fibrillation, which eventually leads to progression to heart failure. Chest X-ray, electrocardiography, echocardiography and colour Doppler ultrasound are usually used for diagnosis, although MRI is the gold standard. Finally, for treatment, pharmacological measures such as beta-blockers or calcium blockers are usually used; otherwise, techniques such as septal reduction therapy with myomectomy or septal ablation with alcohol are usually performed. The implantation of a defibrillator or pacemaker is due to complications that may arise after invasive procedures. New pharmacological therapies are a good option in the case of finding alternatives to invasive procedures.

## FINAL STATEMENT

This review is based on an article by Santiago Vintimilla called “Miocardiopatía Hipertrófica Obstructiva del ventrículo izquierdo”, whose author authorized the translation and rewriting from the Spanish language version to the english language version.

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