

Co-existing subaortic stenosis in a patient with hypertrophic obstructive cardiomyopathy (HOCM) - A rare and interesting finding

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## Abstract

Hypertrophic obstructive cardiomyopathy (HOCM) is an autosomal dominant disorder leading to left ventricular outflow tract obstruction (LVOTO). It can present with chest pain, syncope, breathlessness, or in some cases sudden cardiac death. Primarily, it is diagnosed based on echocardiographic findings but cardiac computed tomography (CT) or cardiac magnetic resonance imaging (MRI) can be helpful in selected cases. In this case report, we discuss a case of a young-aged female patient previously diagnosed as HOCM and presented with chest pain, shortness of breath, and palpitations. Her echocardiography revealed severe asymmetrically hypertrophied left ventricle (LV) with normal function and systolic anterior motion of the mitral valve was present and a subvalvular aortic membrane was also seen. The computed tomography (CT) was also performed showing severe asymmetrical hypertrophied and thickened trileaflet tricommissural aortic valve with no calcification or significant valvular aortic stenosis but there was a subaortic membrane (concentric only sparing anteriorly). The presence of subaortic membrane with HOCM is a rare finding and it can be a diagnostic challenge and untreated cases are susceptible to progressive heart failure and worsening of the symptoms by further increasing left ventricular outflow tract obstruction (LVOTO). A thorough investigation and planning before surgical intervention is required to achieve optimal results.

Keywords: HOCM; SAS; Subaortic Membrane; LVOTO; ASH; SAM

## Background

Hypertrophic obstructive cardiomyopathy (HOCM) is an autosomal dominant disorder associated with the mutation in the genes that encode for the sarcomere proteins. It is defined as cardiac hypertrophy (wall thickness  $\geq 15$  mm, in one or more LV myocardial segments) that is not explained by abnormal loading conditions (e.g. hypertension) and left ventricular obstruction greater than or equal to 30 mm Hg (1). The global prevalence is recorded as 1 in 500 (2). The patient typically presents with exertional chest pain, breathlessness, palpitations, and syncope, while sudden death is the severe manifestation of the disease (3). Though HOCM remains the most common cause of sudden death in athletes, pharmacologic, electrical, and surgical interventions have reduced mortality to 0.5% per year (4). The electrocardiogram (ECG) findings may be non-specific and vary from T wave inversion to findings suggestive of left ventricular hypertrophy (LVH). While the echocardiography is more specific revealing asymmetrical septal hypertrophy (ASH), systolic anterior motion (SAM) of the mitral valve leaflets, left ventricle outflow tract obstruction (LVOTO), and secondary mitral regurgitation (3).

In this case report, we will discuss a patient with hypertrophic obstructive cardiomyopathy (HOCM) who also had subvalvular aortic stenosis (SAS) which is an interesting and rarely reported finding in patients who had HOCM (5). In most cases of subvalvular aortic stenosis (also known as subaortic stenosis), there is a membrane (usually muscular) just beneath the aortic valve causing a fixed obstruction to the blood flow across the left ventricular outflow tract (LVOT) (6). Our case report mentions the diagnostic findings of SAS coexisting with HOCM and describes the treatment options.

## Case

25 years old female patient known case of hypertrophic obstructive cardiomyopathy (HOCM), presented with complaints of chest pain, shortness of breath, and palpitations. The patient had complained of palpitations for many years but it has exacerbated for 3 months. She also had shortness of breath for the last 3 months [New York Heart Association (NHYA) class III] and atypical chest pain. She denied a previous history of syncope, or any chronic medical disorder or any drug history. She also denied any sudden death in her family. On examination, a young age female patient lying on the bed well oriented with time, place, and person. Her blood pressure was 100/70 mmHg, pulse was 70 beats/min and regular, she was afebrile and her respiratory rate was 18/min. On her precordial examination, the apex beat was located at 5<sup>th</sup> intercostal space lateral to the midclavicular line, and double apical impulse was appreciated. On auscultation, S1 and S2 heart sounds were audible of equal intensity and there was ejection systolic murmur (Grade 4+) at the aortic area radiating to the carotid area. On further investigation, her electrocardiograph (figure 1) showed sinus rhythm with left ventricular hypertrophy (LVH) with deep T wave inversions and ST depression depicting LVH with strain pattern. The echocardiography revealed severe asymmetrically hypertrophied left ventricle (LV) with normal function and the systolic anterior motion of the mitral valve. The aortic valve was thickened with no calcification and there was also a subvalvular aortic membrane seen. There was a left ventricle outflow tract (LVOT) dynamic obstruction as well. Specific findings of echocardiography are discussed in table 2. The cardiac computed tomography (CT) was also performed which showed a thickened trileaflet tricommissural aortic valve with no calcification on leaflets and aorta with no significant valvular aortic stenosis. There was severe asymmetrical hypertrophied LV, and diverticulum at mid muscular septum bulging of the membranous part of the interventricular septum (IVS) towards the right ventricle (RV) and a subaortic membrane (concentric only sparing anteriorly) resulting in LVOT obstruction. The basic laboratory workup is mentioned in table 1.

The case of this patient was discussed with the heart team and it was decided to carry out left heart catheterization first and later surgical myomectomy and resection of the subaortic membrane. But the patient requested for discharge and she was kept on medical therapy with beta-blockers and advised for close follow up.

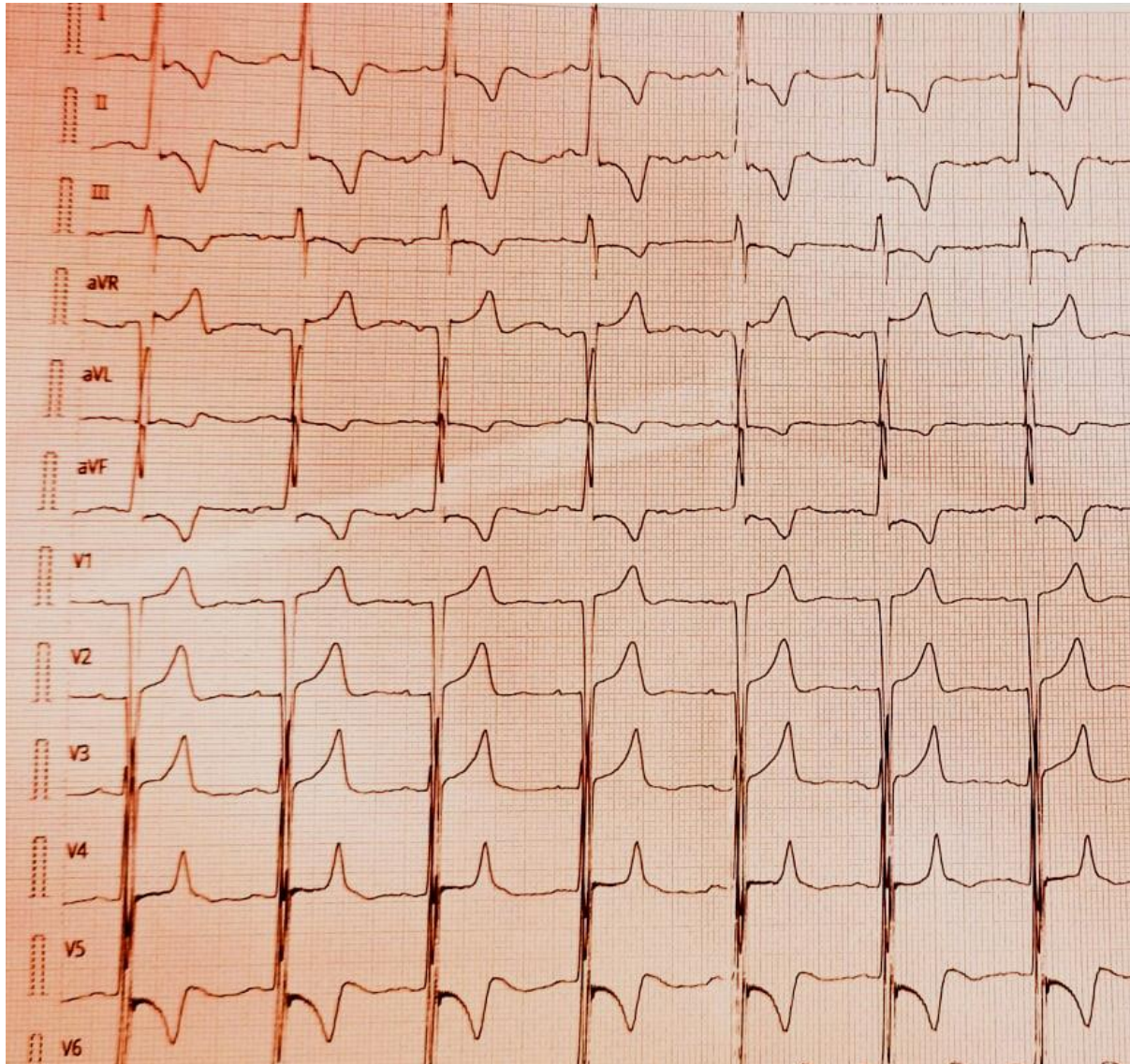


Figure 1: Electrocardiogram (ECG) of the patient: It shows sinus rhythm with left ventricular hypertrophy (LVH) with deep T wave inversions and ST depression depicting LVH with strain pattern

Table 1: Basic Laboratory work-up

Laboratory Investigations	Results	Normal Values
Hb	12.8	14.0-17.4 g/dl
MCV	78.2	76.5-96 fl
TLC	12.3	5.00-10.00 x 10 <sup>9</sup> /L
Neutrophils	62%	50-75%
Lymphocytes	32%	25-40%
Platelets	305	140-400 x 10 <sup>9</sup> /L
Urea	20	10-50 mg/dl
Cr	0.6	0.5-1.2
Na	137	136-149 mEq/L
K	4.2	3.50-5.50 mEq/L
Troponin I	2.67	0.0572 ng/ml
PT	12.8	9.3-14.0 seconds
INR	1.1	0.8-1.2
ALT	26	10-35 U/L
AST	21	0-31 U/L
GGT	22	0-31 U/L
ALP	71	46-116 U/L

Acronyms: Hb: hemoglobin; MCV: mean corpuscular volume; TLC: total leukocyte count; Cr: Creatinine; Na: Sodium; K: Potassium; PT: Prothrombin Time; INR: international normalization ratio; ALT: Alanine Transaminase; AST: Aspartate Aminotransferase; GGT: Gamma-Glutamyltransferase; ALP: Alkaline Phosphatase

Table 2: Specific dimensions given in echocardiography

Septal thickness (normal 11 mm)	24 mm
Posterior wall thickness (normal <11 mm)	21 mm
LVEF (>55%)	65%
Aorta (normal <40 mm)	28 mm
LA (normal <40 mm)	30 mm
LV systolic (normal <34 mm)	26 mm
RV (normal <25 mm)	31 mm
LVOT PG	140 mm Hg

Acronyms: mm: millimeter; LVEF: Left ventricle ejection fraction; LA: Left atrium; LV: Left ventricle; RV: Right ventricle; LVOT PG: left ventricle outflow tract pressure gradient



## Discussion

The patient's clinical presentation and diagnostic findings like asymmetrical septal hypertrophy (ASH) of 24 mm, systolic anterior motion (SAM) of the mitral valve, and left ventricular outflow tract obstruction (LVOT) dynamic obstruction on echocardiography and left ventricular hypertrophy (LVH) with strain pattern on ECG were consistent with hypertrophic obstructive cardiomyopathy (HOCM). Subaortic membrane was noticed on echocardiography as well as in cardiac computed tomography (CT). The subvalvular aortic membrane is an uncommon finding and it can mimic or coexist in patients with HOCM (7). One case report mentioned, the subaortic membrane mimicked valvular aortic stenosis (AS) in which a middle-aged woman with heart failure who was referred for consideration of aortic valve replacement but echocardiography revealed a subaortic membrane with an increased pressure gradient, which resolved after surgical resection (8). Thus, it is suggested that evidence of LVOT obstruction in the absence of systolic anterior motion or valvular aortic stenosis should be considered as a potential subaortic membrane until proven otherwise (7). The subaortic membrane may remain an under-appreciated mechanism of obstruction in patients with HOCM and it increases the susceptibility of progressive heart failure and worsening of the symptoms by further increasing left ventricular outflow tract obstruction (LVOTO) (9).

The patients with LVOTO can be treated with non-vasodilating beta-blockers or calcium channel blockers (if former not tolerated or ineffective). Patients who remain symptomatic with LVOTO >50 mmHg, NYHA class III-IV, and/or recurrent exertional syncope despite maximum tolerated medical therapy should be considered for invasive treatment. The main invasive methods for relieving LVOTO are surgical myomectomy (Morrow procedure) or septal alcohol ablation (3). The subaortic membrane may require surgical myomectomy for definitive relief as percutaneous alcohol ablation would be ineffective (9). However, there is also evidence of the recurrence of the subaortic membrane after surgical intervention and the need for reoperation in various cases (10). In our case report, the patient was advised for surgical myomectomy and resection of the membrane to decrease pressure gradient across LVOT but the patient requested for discharge on request and opted for pharmacological therapy. The implantable cardiac defibrillator (ICD) device can be advised after sudden death risk assessment in HOCM patients and ICD may be considered if a 5-year risk is 4–6% while ICD should be considered if the 5-year risk is >6%. (3).

Thus, diagnosing the subaortic membrane in patients with HOCM requires a high index of suspicion; undiagnosed cases may result in progressive worsening of the symptoms and severe consequences. Echocardiography and cardiac computed tomography (CT) remain helpful diagnostic modalities and in selected cases, cardiac computed tomography (CT) and cardiac magnetic resonance imaging (MRI) are also useful. Preoperative planning and identification of this rare entity are important for optimal results (11).

## Conclusion

The subaortic membrane also is known as subaortic stenosis (SAS) is a rare entity that can be isolated or co-exist with HOCM and cause LVOT obstruction. A patient without any systolic anterior motion of mitral valve or valvular aortic stenosis but having LVOT obstruction should be suspected for subaortic membrane until proven otherwise. Thus, severe consequences can be avoided by early identification and treating the cause. Our case report enlightens a clinical scenario in which subaortic membrane co-exists in a patient with HOCM and it also shares the diagnostic findings of imaging modalities and illustrates the management options for this patient.

## Conflicts of Interest

The authors declare that they have no conflicts of interest.

Note: Informed patient consent form (in English and Urdu) attached in supplementary file

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