Adult Presentation of Subaortic Stenosis: Another Great Hypertrophic Cardiomyopathy Mimic.

Ataul Qureshi MD
Lehigh Valley Health Network, Ataul.Qureshi@lvhn.org

Stephen Awuor MD
Lehigh Valley Health Network, Stephen.Awuor@lvhn.org

Matthew W. Martinez MD
Lehigh Valley Health Network, matthew_w.martinez@lvhn.org

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Adult Presentation of Subaortic Stenosis: Another Great Hypertrophic Cardiomyopathy Mimic

Ataul Qureshi, MD *, Stephen Awuor, MD, Mathew Martinez, MD

Lehigh Valley Health Network, Allentown, PA, USA

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Subaortic stenosis (SAS) is a rare entity in adults with an unclear aetiology and variable clinical presentations and outcomes. SAS typically tends to occur in the first decade of life either as an isolated lesion or in association with other congenital heart diseases. The clinical presentation of SAS can closely mimic hypertrophic cardiomyopathy (HCM) with obstructive physiology.

We present two cases of SAS in adults that were initially presumed to be HCM. The patients were in their late forties and were referred to HCM clinic for further evaluation. Careful review of the transthoracic echocardiogram was indicative for the presence of possible subaortic membrane. These patients underwent subsequent imaging studies that completed the diagnosis for SAS and thereafter had successful surgical resection of the subaortic membrane.

Subaortic stenosis remains a rare and clinically challenging diagnosis in the adult population. Often a combination of imaging modalities is needed to distinguish SAS from HCM with obstruction. It is critical to make the appropriate diagnosis as the treatment options are vastly different from the SAS and HCM with obstruction as well as the implications of a diagnosis of HCM with regards to risk of sudden death and family screening.

Keywords Subaortic stenosis (SAS) • Hypertrophic cardiomyopathy (HCM) • LVOT gradient • Systolic anterior motion (SAM) • Discrete subaortic stenosis • Aortic insufficiency

Introduction

Subaortic stenosis (SAS) is a rare entity in adults with an unclear aetiology and variable clinical presentations/outcomes. SAS typically tends to occur in the first decade of life either as an isolated lesion or in association with other congenital heart diseases. SAS usually follows a progressive course characterised by significant left ventricular outflow tract obstruction (LVOT), left ventricular hypertrophy and aortic valve destruction with subsequent regurgitation. In some instances, there is concomitant systolic anterior motion (SAM) of the mitral valve much like that seen in hypertrophic cardiomyopathy with obstruction. Our cases describe a subaortic membrane in two patients who had initially been thought to have hypertrophic cardiomyopathy with obstruction.

Case report

The first patient is a 47 year-old female with a lifelong murmur referred to Lehigh Valley Health Network Hypertrophic Cardiomyopathy program for evaluation. She described progressive symptoms of exertional chest discomfort, dyspnoea and leg cramps over the preceding several months. The symptoms were new and progressive. She did not have a history of coronary artery disease, hypertension, diabetes mellitus or dyslipidaemia. Her physical examination revealed a bifid apical impulse, grade 3/6 SEM at rest that accentuated slightly with Valsalva manoeuver. She had been evaluated by her cardiologist with a transthoracic echo that was concerning for hypertrophic cardiomyopathy with severe systolic anterior motion (SAM) of the mitral valve and

*Corresponding author at: Lehigh Valley Hospital, 1250 S Cedar Crest Blvd, Allentown, PA 18103, United States. Tel.: +13125324947,
Email: ataul.qureshi@lvhn.org

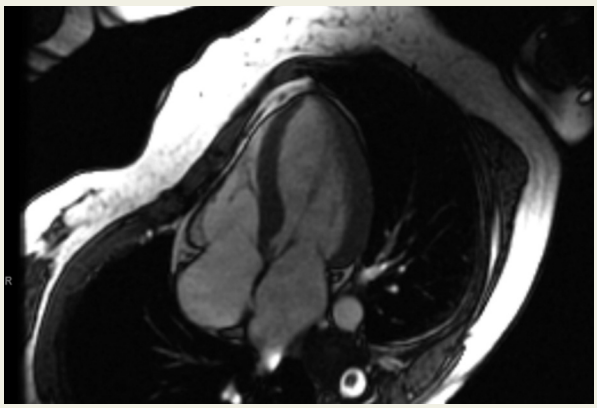

Figure 1 MRI image showing subaortic membrane at the junction of the septal leaflet of the mitral valve and interventricular septum.

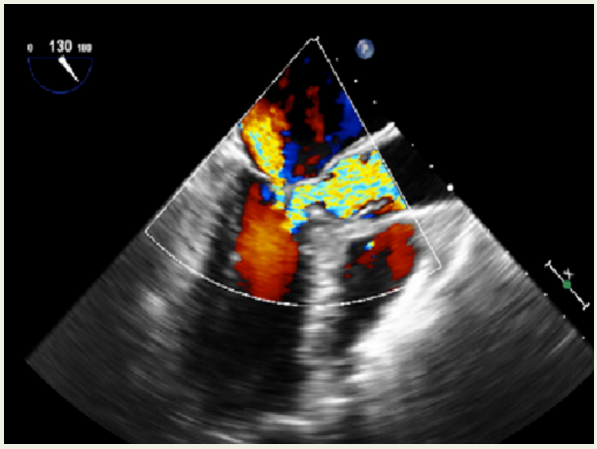

Figure 2 Colour flow acceleration at the level of the subaortic membrane.

a resting LVOT peak/mean gradient of > 90 mmHg. Her symptoms at the time were attributed to hypertrophic cardiomyopathy with resting obstruction. She had been treated with a beta blocker with a resting average heart rate of about 60. She did not have associated syncope, dizziness or palpitations. Further review of her transthoracic echocardiography revealed mild aortic insufficiency and was noted for the presence of possible subaortic membrane (Figure 1).

Further imaging with cardiac MRI revealed a subaortic membrane with flow acceleration in the LVOT and a mildly thickened LV septum. To further define the anatomy the patient underwent a transoesophageal echocardiography (TEE) that demonstrated a subaortic membrane 1.7 cm from the aortic valve annulus severe fixed outflow tract obstruction and aortic regurgitation (Figures 2 and 3). The peak gradient across the subaortic membrane was 100 mmHg (Figure 4). In consideration of the patient's clinical status and her echocardiography findings, she was diagnosed with Discrete Membranous Subaortic Stenosis and recommended

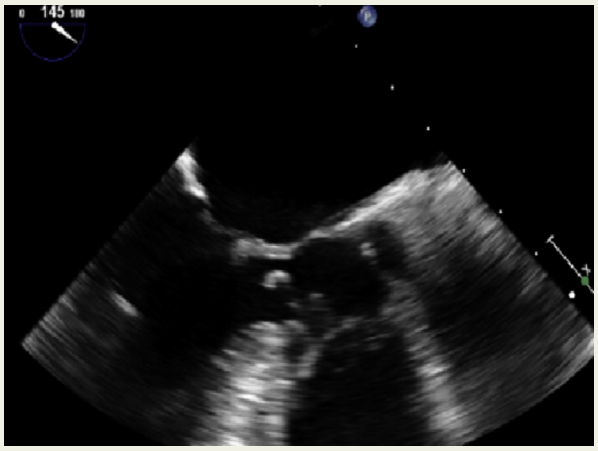

Figure 3 Subaortic membrane seen on the TEE.

for surgical intervention. She underwent surgical resection of the membrane with preservation of her aortic valve without complications. Her symptoms improved at her six-week follow-up visit.

The second patient is 49 year-old female who was also referred for evaluation of a significant murmur with concern of hypertrophic cardiomyopathy. She did not have any chest pain, palpitations, dyspnoea, presyncope or syncope. Exam was significant for a 3/6 systolic ejection murmur with radiation to the carotids, without any change with provocative manoeuvers. She underwent an echocardiogram that showed a resting LVOT gradient of 55 mmHg and mild concentric hypertrophy and mild aortic insufficiency. A subsequent TEE showed discrete subaortic membrane without SAM and there was mild aortic insufficiency. Patient subsequently underwent surgery for the resection of subaortic membrane without any complications and was doing well at the two months post-op follow-up.

Discussion

Discrete subaortic stenosis is a form of left ventricular outflow tract obstruction that usually presents in the first 10 years of life [6,7] as a progressive disease with an overall prevalence of 6.5-20% [1–4]. In up to 10% of patients, the subaortic stenosis is diffuse forming a narrow, tunnel-shaped LVOT [5]. Subaortic stenosis may be associated with the presence of other congenital anomalies especially membranous VSD that can occur in up to 65% of the cases [8]. Other associated congenital anomalies can include bicuspid aortic valve, coarctation of the aorta, and patent ductus arteriosus [9,10]. The exact aetiology of subaortic stenosis is unknown. One of the theories explaining acquired subaortic stenosis proposes the presence of an underlying morphology that is prone to increased turbulence at the LVOT, which in turn increases septal shear stress, producing local reactive cell proliferation leading to the development of the subaortic membrane [11–18].

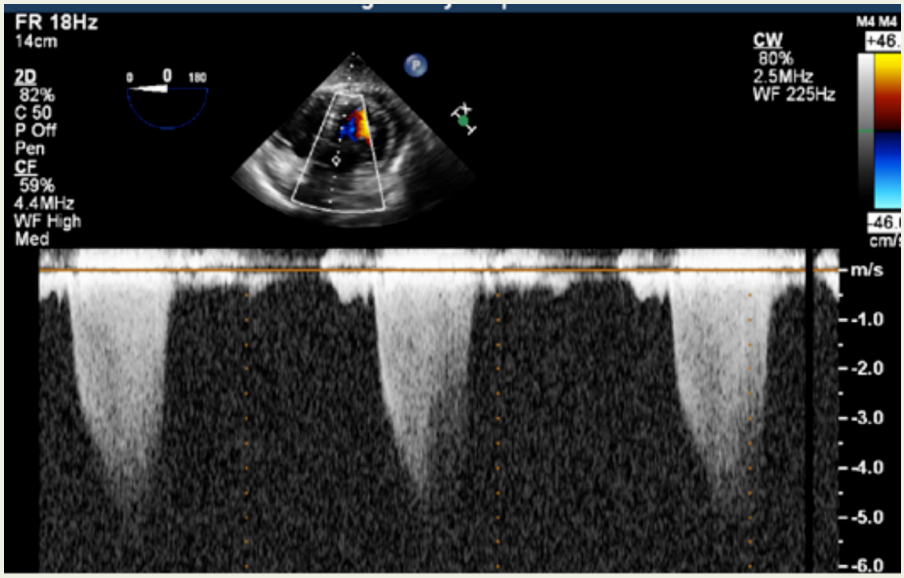

Figure 4 Transgastric TEE view at 0 degrees showing an LVOT gradient of 5 m/sec.

Patients with SAS can range from asymptomatic to varying degrees of symptoms including dyspnoea at rest or with exertion, palpitations, chest pain or syncope. Physical examination may reveal a systolic ejection murmur loudest at the left mid-sternal border radiating to the upper sternal border. Presence of a systolic click may indicate that the lesion is valvular rather than subvalvular. A concurrent diastolic murmur may indicate aortic regurgitation that is commonly associated with SAS [19].

SAS generally tends to progress slowly with an annual increase in the gradient across the stenotic lesion of 2.3 mmHg per year [10]. An intervention is indicated for SAS with mean LVOT pressure gradient greater than 50 mmHg and/or left ventricular systolic dysfunction in order to prevent progression to LVOT obstruction and worsening aortic regurgitation [20–25]. Surgery is the preferred treatment for severe and symptomatic SAS [28–30]. The recurrence of subaortic stenosis varies depending on the type of the preoperative lesion. Valeske et al. reported a recurrence rate after surgery of 16% for membranous SAS and 46% for complex tunnel obstructive lesions [26]

S De Lezo et al. studied the long term outcome of patients with isolated thin membrane subaortic stenosis treated with balloon dilation. They found that 77% of patients had a sustained relief at subsequent follow-ups without restenosis, the need for surgery, progression to muscular obstructive disease, or an increase in the degree of aortic regurgitation. Balloon dilation should be limited to patients with thin membranes [31]. The optimum time for intervention remains to be defined.

The clinical picture of SAS can mimic aortic stenosis as well as hypertrophic cardiomyopathy with obstruction in cases where there may be accompanying asymmetric septal hypertrophy. The clinical distinction between the two entities is important in terms of patient management. A patient with SAS will have a very different evaluation and follow-up compared to those with HCM. In those patients with HCM, one must consider important additional features of HCM including assessment for the risk of sudden death, genetic considerations for family members and associated conditions with HCM such atrial and ventricular arrhythmias. Our patients were initially suspected to have hypertrophic cardiomyopathy with resting obstructive physiology on the basis of clinical presentation, physical findings and SAM of the mitral valve in one of the patients. A detailed analysis of TTE revealed a suspicious membrane in the LVOT which was confirmed by further imaging. Choi et al. presented a similar case of a 58 year-old female with congestive heart failure from SAS associated with SAM. The patient eventually underwent resection of the subaortic membrane and mitral valve replacement with complete resolution of her left ventricular outflow tract obstruction [27]. Thankfully our patients did not require an aortic or mitral valve replacement.

Conclusion

Subaortic stenosis remains a rare and clinically challenging diagnosis in the adult population. Its clinical presentation can closely mimic hypertrophic cardiomyopathy with obstructive physiology. A diagnosis requires a heightened suspicion in patients with LVOT obstruction with associated aortic regurgitation. Often a combination of imaging modalities is needed to distinguish SAS from HCM with obstruction. It is critical to make the appropriate diagnosis as the treatment options are vastly different from the SAS and HCM with obstruction as well the implications of a diagnosis of HCM with regards to risk of sudden death and family screening.

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