When There is More Than Meets the Eye... Should you Trust Your Ears?

Sean Hatzigeorgiou¹, Lauren Basmadjian¹

Affiliations

1. Division of Cardiology, Jewish General Hospital, McGill University, Montreal, Quebec, Canada

Clinical Presentation:

A 31 year-old male with no past medical history presented to hospital with 4 days of upper respiratory symptoms and fever. Physical examination revealed a harsh 3/6 holosystolic murmur at the left sternal border, as well as a 2/6 early diastolic murmur along the right upper sternal border. Transthoracic echocardiogram (TTE) was performed.

Imaging:

TTE revealed eccentric left ventricular hypertrophy with severe cavity dilation and preserved systolic function. A mobile echo density was visualized in the left ventricular outflow tract below the right coronary cusp, with associated aortic root dilation and severe aortic regurgitation (AR). Mild-to-moderate eccentric posteriorly directed mitral regurgitation (MR) was also described.

A transesophageal echocardiogram (TEE) was performed to clarify discordance between physical exam and TTE findings (i.e. unexplained harsh systolic murmur) and to exclude infective endocarditis (IE). A moderately restrictive peri-membranous ventricular septal defect (VSD) with left-to-right shunt was identified. Moderate-to-severe aortic root dilation with moderate AR was noted. Eccentric MR appeared moderate-to-severe. There were no vegetations.

Discussion:

VSDs are common accounting for 10% of congenital heart disease in adults. Most cases are diagnosed in infancy; however, patients may go undetected until adulthood. VSDs can cause left-sided cavity dilation +/- LV dysfunction; pulmonary arterial hypertension; valvular regurgitation, most often due to cusp prolapse into the defect; double chambered right ventricle; and IE. TTE is the most valuable diagnostic tool for VSDs with a sensitivity of 95% and the ability to identify the location, size and consequences of VSDs.

For these reasons, it is important for echocardiographers and sonographers to maintain a high degree of suspicion for VSD in patients presenting with systolic murmurs and/or otherwise unexplained TTE findings associated with VSDs (i.e. LV dilation such as in our patient). This case also highlights the importance of thorough sonographic technique with full sweeps and off-axis images to ensure that critical diagnoses are not missed.

It also underscores that physical examination findings remain essential for diagnosis and guidance of auxiliary testing. In cases of discrepant findings, complimentary imaging such as TEE should be pursued. This modality is also helpful for detecting suspected complications of VSD such as leaflet prolapse and IE.



Figure 1: Panel A: Transthoracic Parasternal long-axis (PLAX) view demonstrating a dilated LV cavity with eccentric LV hypertrophy. **Panel B:** Transthoracic PLAX view depicting severe AR. **Panel C:** Transesophageal modified 4-chamber view with and without colour doppler demonstrating a perimembranous VSD with left-to-right shunt. **Panel D:** Transesophageal 45-degree short-axis view with doppler of the LVOT re-demonstrating a perimembranous VSD with left-to-right shunt.