Incidentally detected Gerbode defect in a patient of type 2 diabetes mellitus

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DESCRIPTION

A 44-year-old woman, known case of type 2 diabetes mellitus with poor glycaemic control (glycated haemoglobin 10.8%), was hospitalised for continuous glucose monitoring (CGM). On enquiring retrospectively during her hospital stay, she also reported a history of effort intolerance, palpitation and exertional dyspnoea (New York Heart Association class II) for 5 years, which she neglected. There were no reports of angina, orthopnoea, paroxysmal nocturnal dyspnoea and pedal swelling. Also there was no history of valvular heart disease, systemic hypertension or blood transfusions. Considering and taking note of all these symptoms, in addition to CGM, patient was thoroughly examined and investigated.

The cardiovascular examination shows normal precordium with apical impulse in left sixth intercostal space (ICS), no palpable thrill, resonant second ICS and no parasternal heave. On auscultation grade III pansystolic murmur was heard over the entire precordium, with maximum intensity along lower left sternal border, without any radiation to axilla. S1 and S2 could not be appreciated in mitral and tricuspid area due to murmur. S2 was heard normally over pulmonary area, with no evidence of loud P2. On dynamic auscultation, murmur intensity increased with inspiration, squatting from standing position and handgrip manoeuvre, while reduced after standing and valsalva manoeuvre. There was no evidence of cyanosis, clubbing and tender hepatomegaly. The jugular venous pressure was 7 cm H₂O.

The ECG shows sinus rhythm with no evidence of left/right ventricular hypertrophy. Chest roentgenogram did not show any evidence of cardiomegaly or dilated pulmonary artery. The transthoracic echocardiography (TTE) revealed normal left ventricular ejection fraction, mildly dilated right atrium (RA) and ~6–7 mm-sized ventricular septal defect (VSD) connecting left ventricle (LV) to RA with left-to-right shunt through tricuspid valve (TV), suggestive of Gerbode defect. Transoesophageal echocardiography (TOE) confirmed the findings of TTE (figures 1 and 2) showing turbulent flow through the defect (online supplementary videos 1 and 2), with a pressure gradient of ~110–120 mm Hg. She was planned for surgical repair of the defect.

Gerbode defect is a rare type of VSD, with direct communication between LV and RA, accounting for 0.08% of intracardiac shunts and <1% of all congenital cardiac defects.^{1–3} It was first described in 1838⁴ and named after

Figure 1 Transoesophageal echocardiography (TOE) midoesophageal (ME 0 degree)—four-chamber view showing communication between the left ventricle to the right atrium through the tricuspid valve.

Gerbode *et al*⁵ successfully performed surgery on five patients with this defect in 1958. The supravulvar (direct shunt), infravalvular (indirect left-to-right shunt with associated TR) and mixed are three major types of the defect. The clinical presentations include asymptomatic cases, chest pain, shortness of breath, lower extremity oedema, severe right and/or left heart failure with high mortality; based on size, volume and duration of the shunt.^{1,6} Most patients have a systolic murmur, heard over the entire precordium.^{1,7}

TOE serves as the most sensitive modality for detecting LV-RA shunts.⁷ The infra-valvular defect shows unidirectional shunt flow across the TV; thereby differentiating it from Eisenmenger VSD with Tricuspid Regurgitation (TR) exhibiting bidirectional shunt flow.⁸ Gerbode defect typically shows a systolic left-to-right shunt that needs to be differentiated from ruptured sinus of valsalva aneurysms, which also shows shunting of blood during diastole due to diastolic gradient established between aorta and RA.⁸ The surgical management of the defect is done based on the severity

Figure 2 Transoesophageal echocardiography (TOE) midoesophageal (ME 0 degree)—four-chamber view with colour doppler showing turbulent flow from the left ventricle to the right atrium through the tricuspid valve.

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Learning points

- Gerbode defect is a rare cardiac anomaly and is a type of ventricular septal defect (VSD), with direct communication between left ventricle and right atrium.
- Patients with Gerbode mostly have a systolic murmur, heard over the entire precordium.
- The clinical presentation, physical examination and findings of echocardiography serve as the important aid to the differential diagnosis of infravalvular Gerbode defect from Eisenmenger VSD with tricuspid regurgitation.

of symptoms, clinical judgement and presence of comorbid conditions.

The detailed history considering all the symptoms that the patients present with and the physical examination carries utmost importance even in the era of advanced investigations and diagnostics. If carefully considered, they serve as valuable guide to use these advancements for timely diagnosis and planning intervention in patient care (as in our case), thereby halting disease progress and preventing complications.

Contributors RR conceived the idea of the case report. RR, RN and MA provided the scientific guidance for the workup of the case. MR has worked with the case

investigations, acquisition and interpretation of the data. MR and RN have worked for drafting and reporting the manuscript.

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