Infravalvular type of Gerbode defect: a rare cardiac anomaly

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DESCRIPTION

A Gerbode defect is a rare type of ventricular septal defect (VSD), classically described as a direct communication between left ventricle (LV) and right atrium (RA).¹ However, it may be

supravalvular (direct shunt), infravalvular (indirect shunt) or mixed. The infravalvular type is a VSD with a left-to-right shunt at the ventricular level, associated with tricuspid regurgitation (TR), which directs the high-velocity shunt into RA.^{2 3} A 43-year-old woman presented with symptoms of dyspnoea and palpitations. Clinical examination revealed a pansystolic murmur and grade 2 parasternal heave. ECG depicted biventricular hypertrophy along with biatrial enlargement. Echocardiogram showed a 9 mm perimembranous VSD with left-to-right shunt (figure 1). Careful observation revealed two separate systolic tricuspid regurgitant jets from right ventricle (RV) into RA, with different timings in systole (figure 2 and video 1). The first was the high-velocity VSD jet, which was directed into the RA through the tricuspid valve (TV) with a high-pressure gradient of 120 mm Hg across it (figure 3). The second was the mild TR jet. Finding two separate systolic jets across TV supported the diagnosis of infravalvular Gerbode defect and ruled out Eisenmenger VSD with TR. On cardiac catheterisation, oximetry revealed a step up in oxygen saturation of 20% at the RA level. Pulmonary artery pressure was

Figure 1 Echocardiogram, apical four-chamber view, demonstrating a perimembranous VSD (arrow) with shunt from LV to RV directed to RA through TR jet. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; TR, tricuspid regurgitation; VSD, ventricular septal defect.

Figure 2 Echocardiogram, modified parasternal short-axis view, demonstrating two separate TR jets: (A) first a large systolic regurgitant jet across the tricuspid valve, suggesting indirect shunting of blood from LV to RA; then (B) a second small systolic regurgitant jet across the tricuspid valve suggesting true TR. LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; TR, tricuspid regurgitation; VSD, ventricular septal defect.

Video 1 Echocardiogram, apical four-chamber view, demonstrating a perimembranous ventricular septal defect with two systolic regurgitant jets across the tricuspid valve from right ventricle into right atrium.

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Figure 3 Echocardiogram demonstrating high jet velocity and high-pressure gradient across the regurgitant jet from right ventricle into right atrium.

Figure 4 (A–D) Successive frames of left ventricle (LV) angiogram showing a subaortic ventricular septal defect (VSD) filling both right ventricle (RV) and right atrium (RA). Note that the initial VSD jet is directed preferentially towards RA (B and C) and then full opacification of both RA and RV occurs simultaneously (D).

Video 2 Left ventricle angiogram showing a subaortic ventricular septal defect with preferential filling of right atrium earlier than right ventricle.

114/39 mm Hg (mean 54). LV angiogram showed a subaortic VSD with preferential filling of RA earlier than RV (figure 4 and video 2). The ratio of pulmonary vascular resistance to systemic vascular resistance was 0.08. The patient underwent successful surgical repair of the defect.

Competing interests None declared.

Patient consent Obtained.

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Learning points

- Gerbode defect (left ventricle–right atrium shunt) is a rare cardiac anomaly that can be supraventricular, infravalvular or mixed, depending on the position of the defect in relation to the tricuspid valve.
- Right atrial enlargement in a case of ventricular septal defect (VSD) should raise the possibility of Gerbode defect.
- The finding of two separate tricuspid regurgitation (TR) jets with different timings in systole helps in differentiating an infravalvular type of Gerbode defect from Eisenmenger VSD with TR.

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