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CASE REPORT
Cardiac imaging

Gerbode defect following surgical mitral valve replacement and tricuspid valve repair: a case report

Rebecca H. Haraf [1], Mohamad Karnib[2], Chantal El Amm[2], Sarah Plummer[3], Martin Bocks[3], and Ellen M. Sabik[2]*

[1]Department of Internal Medicine, University Hospitals Cleveland Medical Center/Case Western Reserve University School of Medicine, 11100 Euclid Avenue, Cleveland, OH, 44106, USA; [2]Division of Cardiovascular Medicine, Department of Internal Medicine, University Hospitals Cleveland Medical Center/Case Western Reserve University School of Medicine, 11100 Euclid Avenue, Cleveland, OH, 44106, USA; and [3]Department of Pediatrics, Division of Pediatric Cardiology, University Hospitals Rainbow Babies & Children's Hospital/Case Western Reserve University School of Medicine, 11100 Euclid Avenue, Cleveland, OH, 44106, USA

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Background Gerbode defect is a congenital or acquired communication between the left ventricle and right atrium. While the defect is becoming a more well-recognized complication of cardiac surgery, it presents a diagnostic and therapeutic challenge for providers. This case highlights the predisposing factors and imaging features that may assist in the diagnosis of Gerbode defect, as well as potential approaches to treatment.

Case summary We report a patient with severe mitral stenosis as a result of remote mediastinal radiation who underwent extensive decalcification during surgical mitral valve replacement and tricuspid valve repair. Following the procedure, he developed progressive heart failure refractory to medical management. Extensive workup ultimately led to the diagnosis of iatrogenic acquired Gerbode defect. Close collaboration between adult cardiology, cardiothoracic surgery, and the congenital cardiology services led to an optimal treatment plan involving percutaneous closure of the defect.

Discussion Gerbode defect is a rare complication of invasive procedures involving the interventricular septum or its nearby structures. An understanding of the key echocardiographic features will aid providers in timely diagnosis. Percutaneous repair should be strongly considered for patients who may be poor surgical candidates.

Keywords Radiation heart disease • Transcatheter ventricular septal defect (VSD) closure • Acute heart failure • Mitral valve • Tricuspid valve • Gerbode defect • Case report

Learning points

- Gerbode defect is a high membranous ventricular septal defect leading to abnormal communication between the left ventricle and the right atrium.
- Severe calcific valve disease, mediastinal radiation, and cardiac surgery involving the membranous septum may predispose to the development of iatrogenic Gerbode defect.
- Gerbode defect can present a diagnostic challenge, but high clinical suspicion in at-risk patients and understanding of the key echocardiographic features can lead to prompt diagnosis and treatment.
- A percutaneous approach to repair of Gerbode defect may be preferable to surgical repair for patients at high risk of surgical morbidity and mortality.

* Corresponding author. Tel: 216-844-5453, Fax: 216-844-8316, Email: Ellen.Sabik@UHhospitals.org
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Introduction

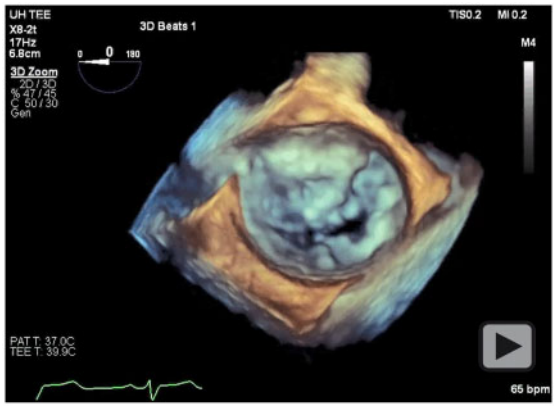
A Gerbode defect is a communication between the left ventricle (LV) and right atrium (RA) due to a high ventricular septal defect. Originally described as a rare congenital defect, recent evidence suggests that the incidence of acquired defects has surpassed congenital defects, with the majority of cases resulting from iatrogenic injury, endocarditis, myocardial infarction in the right coronary artery distribution, or blunt cardiac trauma.[1] With the development of readily available advanced imaging techniques such as a three-dimensional transoesophageal echocardiogram (TOE) and cardiac magnetic resonance, the diagnosis of acquired iatrogenic Gerbode defect has become a known complication of cardiac surgery and percutaneous coronary interventions involving the interventricular septum or its nearby structures.[2]

Timeline

c. 1988	Underwent mediastinal radiation for the treatment of Hodgkin lymphoma
27/9/2012	Referred to cardiothoracic surgery for symptomatic severe radiation-induced aortic stenosis
4/3/2013	Underwent surgical aortic valve replacement with resolution of symptoms and no post-operative complications
8/11/2019	Referred to cardiothoracic surgery for symptomatic severe radiation-induced mitral stenosis with concomitant mitral and tricuspid regurgitation
6/12/2019	Underwent mitral valve replacement and tricuspid valve repair
16/12/2019	Permanent pacemaker placed for post-operative complete heart block
18/12/2019	Developed progressing symptoms of acute decompensated heart failure including weight gain, orthopnoea, and shortness of breath
8/1/2020	Transoesophageal echocardiogram identifies Gerbode defect
11/1/2020	Underwent percutaneous closure of Gerbode defect using septal occluder device
12/1/2020	Symptom resolution
29/2/2020	Discharged home

Case presentation

A 54-year-old male with a remote history of Hodgkin lymphoma treated with chemotherapy and mediastinal radiation was admitted to the hospital to undergo elective surgical mitral valve (MV) replacement and tricuspid valve (TV) repair. He previously had radiation-induced aortic valve stenosis requiring surgical aortic valve replacement with bioprosthetic aortic root in 2013. He was again referred to cardiothoracic surgery in 2019 for shortness of breath, fatigue, and severe pulmonary arterial (PA) hypertension (mean PA pressure 75 mmHg on right heart catheterization) resulting from radiation-induced calcific mitral stenosis, mitral regurgitation (MR) and tricuspid regurgitation (TR). Intraoperatively, there was enormous calcification extending from the interannular fibrosa to the A1, A2, P1, and P3 regions of the mitral valve requiring extensive debridement (*Video 1, Figure 1*). A bioprosthetic MV and TV annuloplasty ring were then successfully inserted with initial improvement in PA pressures by approximately 30% by invasive monitoring. Limited intraoperative TOE post-cardiopulmonary bypass showed preserved ventricular function, trace TR, and trace paravalvular MR in the A1 region where calcification had been heaviest, and debridement caused disruption of the anterior mitral annular structure.



Video 1 Severe calcific mitral stenosis demonstrated on pre-intervention three-dimensional intraoperative transoesophageal echocardiogram.

He was admitted to the intensive care unit for post-operative care and weaned off ventilator and vasopressor support. He was found to have post-surgical complete heart block requiring placement of a permanent pacemaker on post-operative Day 11.

One week later, he developed shortness of breath, orthopnoea, and weight gain. Physical exam was notable for jugular venous distention, bibasilar pulmonary crackles, and peripheral pitting oedema. Despite escalating diuretics, the volume overload progressed with a net weight gain of 16 kg. He developed acute kidney injury, transaminitis, and hypotension requiring inotropic and vasopressor support. Laboratory investigation also revealed worsening haemolytic anaemia (haemoglobin nadir 8.1 g/dL, total bilirubin 43.8 mg/dL, lactate dehydrogenase 2993 U/L, haptoglobin <30 mg/dL, and schistocytes on peripheral smear). Additional testing ruled out adrenal insufficiency (Ante meridiem (AM) cortisol 18.8 µg/dL), hyperthyroidism (thyroid stimulating hormone (TSH) 3.59 mIU/L, free thyroxine 1.25 ng/dL),

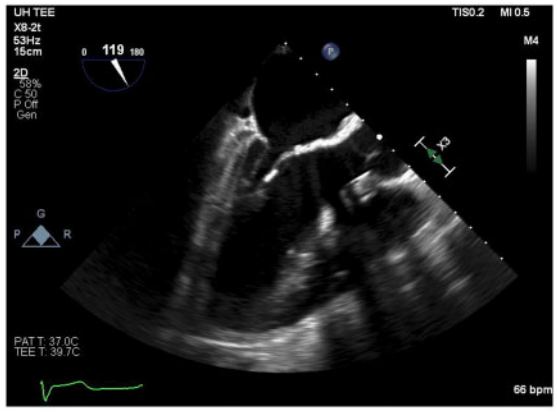


Figure 1 Echocardiogram demonstrating diffuse, radiation-induced calcification extending from the interannular fibrosa to the mitral valve leaflets.

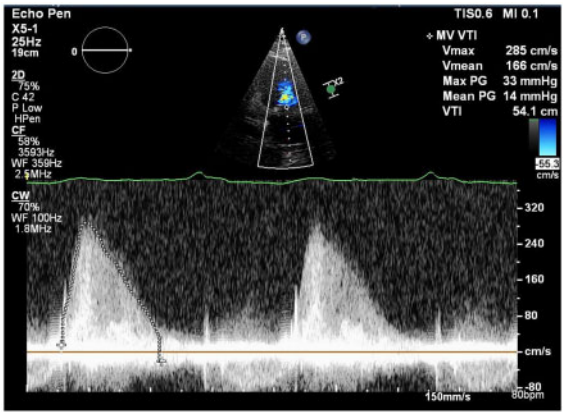


Figure 2 Transthoracic echocardiogram with continuous wave Doppler demonstrating increased transmitral gradient and normal pressure half-time suggestive of occult mitral paravalvular leak with acoustic shadowing by prosthesis.

or arteriovenous fistula at the recent left heart catheterization femoral access site.

Repeat transthoracic echocardiogram (TTE) identified increased transmitral gradient (mean 14 mmHg, peak 33 mmHg) with normal pressure half-time (81 ms), elevated peak velocity (2.8 m/s), elevated velocity Time integral (VTI) ratio (2.92), and decreased indexed effective orifice area (0.4 cm^2/m^2) suggestive of significant occult MR vs. patient-prosthesis mismatch; it also revealed moderate TR and an estimated right ventricular systolic pressure of 80.3 mmHg (*Figure 2*). Due to concern for paravalvular leak causing severe haemolytic anaemia and high-output heart failure, a TOE was performed. Images confirmed a moderate-to-severe medial paravalvular mitral leak and a smaller posterolateral leak without evidence of valvular MR. There was a pacemaker lead crossing through the septal portion of the tricuspid annuloplasty ring with dehiscence and associated moderate TR. Most notably, the TOE unexpectedly identified a large septal defect between the LV and the RA (*Figure 3, Video 2*), making the diagnosis of acquired iatrogenic Gerbode defect.

The Gerbode defect was adjacent to the medial aspect of the MV prosthesis and posterior to the TV apparatus. Because of the significant risk posed by surgical intervention, the decision was made to pursue transcatheter device closure.

The patient subsequently underwent right heart catheterization, which demonstrated mean RA pressure 30 mmHg, right ventricular (RV) pressure 80/8 mmHg with end-diastolic pressure of 28 mmHg, PA pressure 80/30 (52) mmHg, and mean pulmonary capillary wedge pressure 36 mmHg. Intraoperative TOE confirmed a Gerbode defect (6.5 mm × 7.8 mm). The defect was closed via right internal jugular approach using a 12-10mm AMPLATZER Duct Occluder (Abbott, Lake Bluff, IL, USA) with immediate drop in mean RA pressure to 21 mmHg, increase in systolic blood pressure by 30 mmHg, and trace residual shunt (*Video 3*). Because of the hemodynamic significance of the Gerbode defect, as well as the proximity of the defect to the mitral valve prosthesis, the paravalvular mitral leak was not primarily addressed. Incidentally, there was mild improvement of the paravalvular mitral leak following Gerbode closure due to some overlap of the retention disc with this area (*Video 3*).

Following defect closure, there was rapid recovery of renal function, resolution of haemolysis and increased response to diuresis (*Table 1*). On repeat TTE 1 week later, the device remained in appropriate position with insignificant residual shunt. Despite the fact that a mild paravalvular mitral leak persisted with a prosthetic mitral valve mean gradient of 9.5 mmHg, the patient clinically improved, and the decision was made to defer any further attempts at repair. He was discharged after a prolonged hospital course asymptomatic on daily diuretics, including bumetanide, spironolactone, and metolazone. The patient completed his subsequent follow-up visits at an outside facility closer to his home, but 8 months later, he remained well with no hospitalizations following his discharge.

Discussion

Gerbode defect presents a diagnostic challenge both due to its low incidence as well as difficulty in discerning the shunt features on TTE from more common anatomic abnormalities. The typically high-velocity Doppler signal generated by the LV-to-RA shunt in systole may be easily identifiable; however, it is often difficult to distinguish this flow from TR in the setting of confirmed or suspected pulmonary arterial hypertension. This is especially true when surgical implants cause artefacts in the area of interest. Echocardiographic clues that may aid in the diagnosis include atypical jet direction, normal diastolic PA pressure, and the absence of typical RV features of severe PA hypertension (RV hypertrophy and ventricular septal flattening).[3]

In the case presented, multiple TTEs were unable to clearly differentiate flow through the defect from that of high-velocity TR with local bioprosthetic valve shielding. We also speculate that the defect was initially insignificant, thus the intraoperative TOE failed to identify

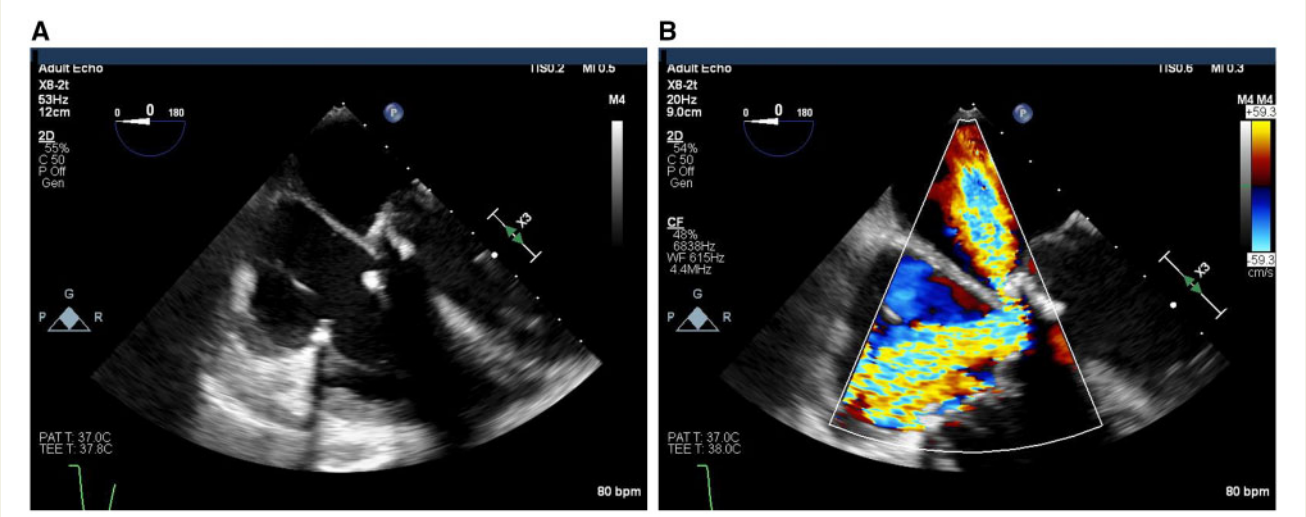
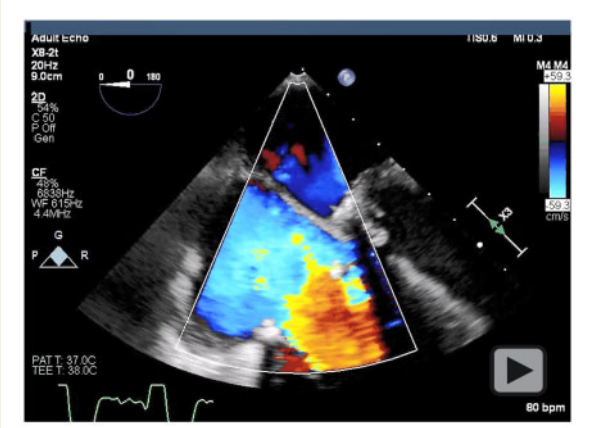
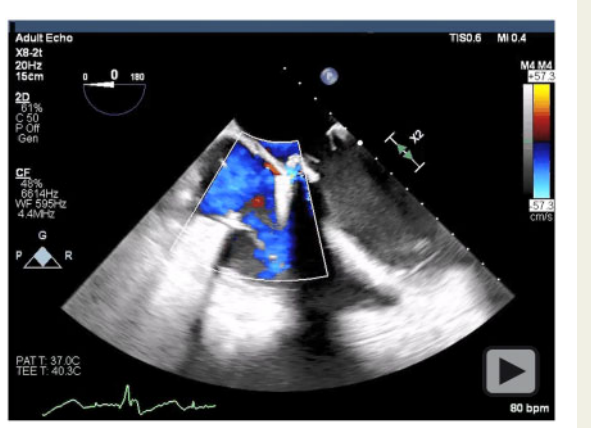


Figure 3 (*A*, *B*) Diagnostic transoesophageal echocardiogram demonstrating a centrally directed tricuspid regurgitant jet and a separate, laterally directed jet through an iatrogenic left ventricle-to-right atrial shunt with concomitant tricuspid annular dehiscence and moderate paravalvular leak in the A1 region of the prosthetic mitral valve.



Video 2 Diagnostic transoesophageal echocardiogram demonstrating a centrally directed tricuspid regurgitant jet and a separate, laterally directed jet through an iatrogenic left ventricle-to-right atrial shunt with concomitant tricuspid annular dehiscence and moderate paravalvular leak in the A1 region of the prosthetic mitral valve.



Video 3 Post-operative transoesophageal echocardiogram demonstrating percutaneous placement of Amplatzer device with near-complete occlusion of left ventricle-to-right atrial flow. Mild residual paravalvular leak persists.

its presence; however, over time, the increased gradient between the LV and the RA made the communication more pronounced. The diagnosis was further confounded by elevated PA diastolic pressure, likely a reflection of residual elevated pulmonary vascular resistance from long-standing MV disease.

It is well-known that mediastinal radiation can lead to a variety of progressive cardiac pathology, including accelerated coronary artery arteriosclerosis, calcific valve disease, restrictive cardiomyopathy, and changes in myocardial thickness.[4] Complications may manifest years after therapy; in one longitudinal study of Hodgkin lymphoma survivors, significant cardiac remodelling was demonstrated to occur as long as 30 years post-treatment.[5] In the case presented, the degree of calcification resulting from prior radiation required extensive debridement, ultimately leading to the formation of the iatrogenic Gerbode defect. Notably, combined tricuspid annuloplasty ring insertion and mitral valve replacement has specifically been identified as a common culprit in the development of Gerbode defect, as attempts at mitral and tricuspid septal leaflet decalcification may easily injure the upper portion of the membranous septum.[1]

Table 1 Pertinent laboratory and hemodynamic data before and after Gerbode defect repair

	Before Gerbode repair	Day of discharge[a]
Haemoglobin	8.1 g/dL	10.4 g/dL
Creatinine	3.29 mg/dL	2.22 mg/dL
Aspartate Aminotransferase (AST)	433 U/L	37 U/L
Alanine aminotransferase (ALT)	124 U/L	16 U/L
Total bilirubin	43.8 mg/dL	3.1 mg/dL
LDH	2993 U/L	615 U/L
Systolic blood pressure	85–115 mmHg	127 mmHg
Mean pulmonary artery pressure	52 mmHg	19 mmHg[b]

[a]Patient was discharged approximately 6 weeks after Gerbode repair.
[b]PA catheterization was performed approximately 6 weeks after Gerbode repair.

Because these patients are often high-risk surgical candidates, reoperation to close an iatrogenic Gerbode defect is often an undesirable option. Thus, percutaneous closure should be considered. Transcatheter device closure is becoming increasingly common for patients that are deemed high risk for reoperation and has been shown to be safe and effective in multiple cases of acquired defects.[6] Although surgical intervention remains the mainstay of therapy, the availability of new devices for structural interventions should prompt consideration of percutaneous approach to repair.[2]

Conclusions

Gerbode defect is a rare but recognized complication of cardiac surgery involving the membranous septum. It presents a diagnostic challenge given its echocardiographic features, requiring high clinical suspicion for at-risk patients in the post-operative period. This case demonstrates that mediastinal radiation can result in extensive valvular calcification and significantly increase surgical morbidity. Close collaboration between adult cardiology, cardiothoracic surgery, and the congenital cardiology services led to the optimal treatment plan for this patient. Treatment is individualized, but percutaneous closure should be strongly considered for those with increased surgical risk given its frequently favourable outcome.

Lead author biography

Rebecca Haraf is a second year Internal Medicine resident at University Hospitals Cleveland Medical Center (UHCMC). Once completing her residency training, she will remain at UHCMC as Chief Resident, after which she intends to apply to Cardiovascular Fellowship.

Supplementary material

Supplementary material is available at *European Heart Journal - Case Reports* online.

Slide sets: A fully edited slide set detailing these cases and suitable for local presentation is available online as Supplementary data.

Consent: The author/s confirm that written consent for submission and publication of this case report including image(s) and associated text has been obtained from the patient in line with COPE guidance.

Conflict of interest: None declared

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