American Journal of Case Reports

e-ISSN 1941-5923
© Am J Case Rep, 2022; 23: e935537
DOI: 10.12659/AJCR.935537

Received: 2021.11.18
Accepted: 2022.03.25
Available online: 2022.03.30
Published: 2022.05.03

An "Unusual" Diverticulated Appearance in Adult Direct Gerbode Defect: A Case Report

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

ABCDEFG **Tri Wisesa Soetisna**

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Financial support: None declared
Conflict of interest: None declared

Patient: Male, 46-year-old
Final Diagnosis: Gerbode VSD • tricuspid regurgitation
Symptoms: Fatigue • shortness of breath
Medication: —
Clinical Procedure: De Vega procedure • defect closure
Specialty: Cardiac Surgery

Objective: Rare disease

Background: Gerbode defect is a defect that communicates the left ventricle (LV) to the right atrium (RA). Although it was originally identified as a congenital defect, it is becoming more common accepted that the lesion has either an iatrogenic or non-iatrogenic origin. In this article, the author presents an unusual diverticulated appearance of direct Gerbode defect in an adult patient without any prior history of cardiac pathology and the surgical technique used, hoping to increase awareness of similar cases.

Case Report: A 46-year-old man presented with worsening shortness of breath and fatigue for 2 years. The patient's medical history was only significant for high blood pressure, with no previous cardiac abnormalities. No other metabolic syndrome was identified in this patient. There was no family history of congenital heart disease. Echocardiography showed a Gerbode defect with left-to-right shunt from LV to RA. The author found a diverticulated lesion around the area of the Gerbode defect intraoperatively, which raised a suspicion that the diverticulum may have initiated the Gerbode defect.

Conclusions: Congenital heart disease is currently one of the most common congenital diseases which require surgical intervention. The vast spectrum of severity in many congenital heart diseases makes the diagnosis and early recognition challenging, so many patients live with the condition undiagnosed until adulthood. Any abnormality which causes left-to-right shunting must be identified as early as possible to prevent further complications such as congestive heart failure.

Keywords: Heart Defects, Congenital • Heart Failure • Heart Septal Defects, Ventricular

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/935537

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Background

One of the rare types of congenital heart defect is the Gerbode defect, a variant of ventricular septal defect (VSD) in which there is a communication between the left ventricle (LV) and right atrium (RA) [1]. First described by Thurnam in 1838, the Gerbode defect was named after a well-known surgeon from Stanford University, Frank Gerbode, who published a case series on the successful surgical repairs of 5 patients in 1958 [2]. Isolated congenital Gerbode defects are rare, while the acquired types of this lesion are more common. The acquired Gerbode defect have iatrogenic, infective, traumatic, or ischemic etiologies [3]. For the congenital type, the pathogenesis is still poorly understood, although NKX2-5, GATA4, and TBX5 genes sequence variations have been implicated in some cases [4].

There are 3 types of communication between LV and RA in the Gerbode defect based on the anatomical location, which are Type 1 (indirect or infravalvular), Type 2 (direct or supravalvular), and Type 3 (intermediate) (**Figure 1**). Type 1 lesions occur when there is an interventricular septal defect, combined with a defect in the tricuspid valve. The direct connection between LV and RA through the superior part of the membranous atrioventricular (AV) septum (supravalvular) is identified in Type 2 lesions. Type 3 intermediate lesions are described when there is a combination of both direct and indirect types of the Gerbode defect [1]. The global incidence of Type 1, 2, and 3 Gerbode defects is 12%, 81%, and 7%, respectively [3]. By following the 2020 Surgical Case Report guidelines, the author presents an unusual appearance of a direct Gerbode defect in an adult patient with no prior history of cardiac pathology [5]. This patient underwent a surgical intervention to repair the defect. The area surrounding the defect had a diverticulated appearance.

Case Report

A 46-year-old male patient was referred to our hospital due to findings of shortness of breath and fatigue identified in the referring hospital. These symptoms had been getting worse since 2019. The patient's medical history was only significant for high blood pressure, with no previous cardiac abnormalities. No other metabolic syndrome was identified in this patient. His surgical history included appendectomy for appendicitis in 2008. There was no family history of congenital heart disease. Transthoracic echocardiography showed a suspicious cardiac anomaly and he was then referred to our hospital for further care.

His vital signs showed blood pressure of 153/77 mmHg, heart rate 73 bpm, respiratory rate 16 x/min, and body temperature 36.7°C. Physical examination findings are significant for shortness of breath on minimal exertion and a pan-systolic murmur. Laboratory investigations revealed a hemoglobin of 15.3 g/dL, hematocrit of 44.5%, white cell count of 10 380/dL, thrombocyte of 286 000/dL, aPTT of 31.5, and INR of 1.01. The patient was taking bisoprolol 10 mg once daily, lisinopril 10 mg once daily, hydrochlorothiazide 12.5 mg once daily, amlodipine 5 mg once daily, and furosemide 40 mg once daily.

A chest X-ray revealed cardiomegaly with a cardio-thoracic ratio of >50% and increased pulmonary vascularity. Electrocardiography showed sinus rhythm. Findings from transthoracic echocardiography identified a normal ejection fraction of 54%, aortic root diameter 37 mm, end-diastolic diameter (EDD) 54.8 mm, end-systolic diameter (ESD) 41.3 mm, left atrium diameter 50 mm, and tricuspid annular plane systolic excursion (TAPSE) 25 mm. There was a 0.5-mm gap at the atrioventricular (AV) septum from LV to RA with left-to-right shunt, consistent with a Gerbode defect (**Figure 2**). Concentric LV hypertrophy with diastolic dysfunction grade I was also observed. Tricuspid regurgitation was difficult to assess due to the same trajectory of the blood flow from the VSD. Histopathological examination of the defect and the measurement of LV to RA flow rate were not performed in this patient. Surgical management was planned on this patient for VSD closure, as well as further intra-operative evaluation to assess tricuspid regurgitation.

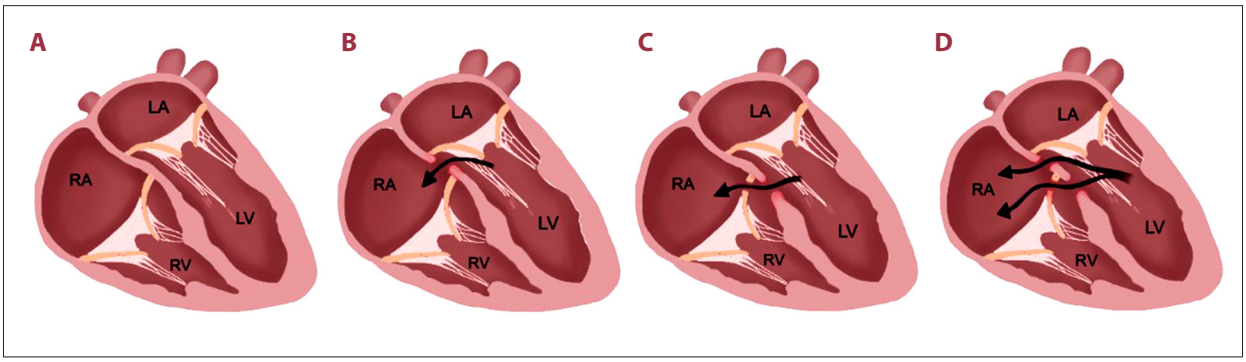

Figure 1. Three types of Gerbode defect. Normal interventricular septum (**A**). Direct Gerbode defect (**B**). Indirect Gerbode defect (**C**). Intermediate Gerbode defect (**D**). Illustration by Soetisna TW (2021).

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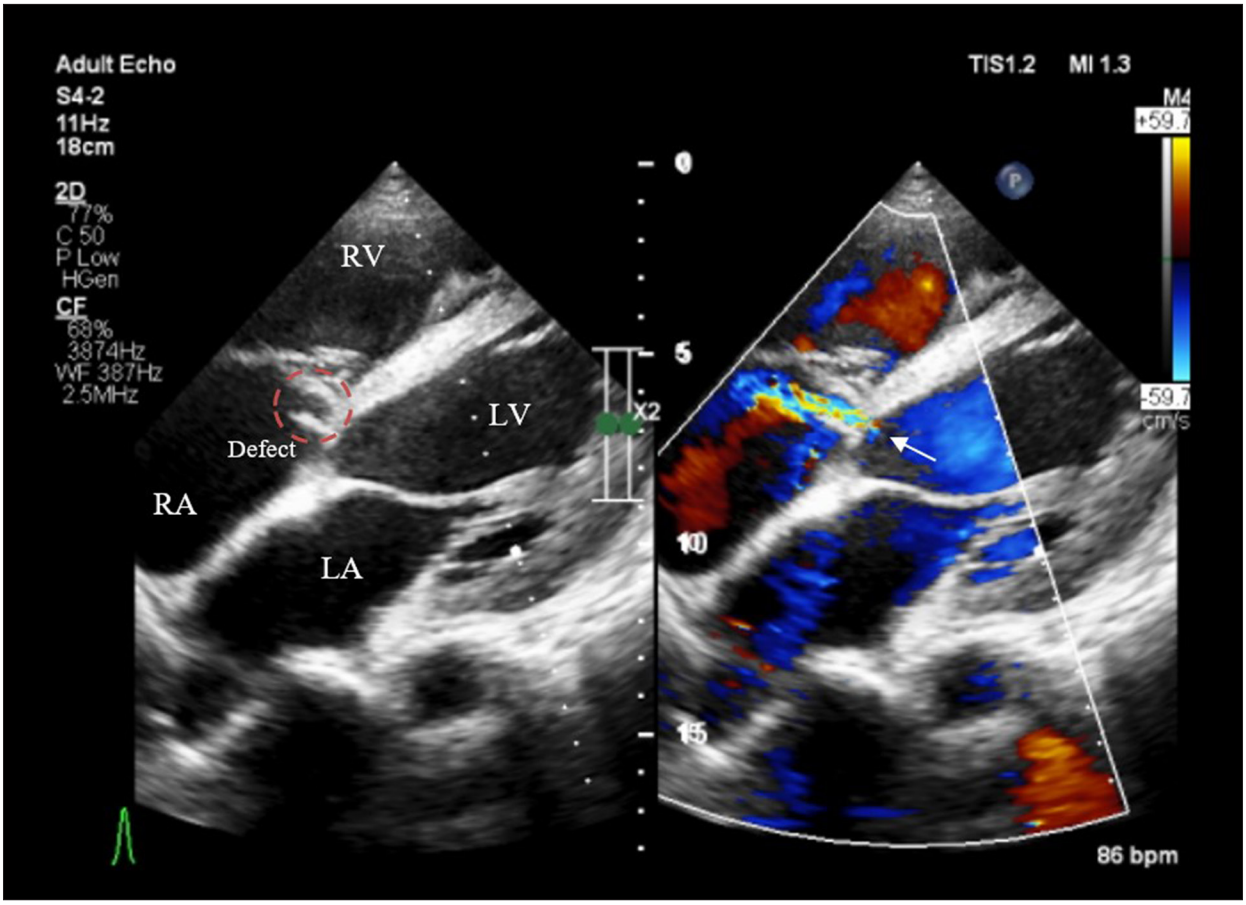

Figure 2. Transthoracic echocardiography. Transthoracic echocardiography revealed 0.5-mm gap (white arrow) at the atrioventricular septum connecting left ventricle with right atrium with left-to-right shunt. RV – right ventricle; RA – right a; LV – left ventricle; LA – left atrium.

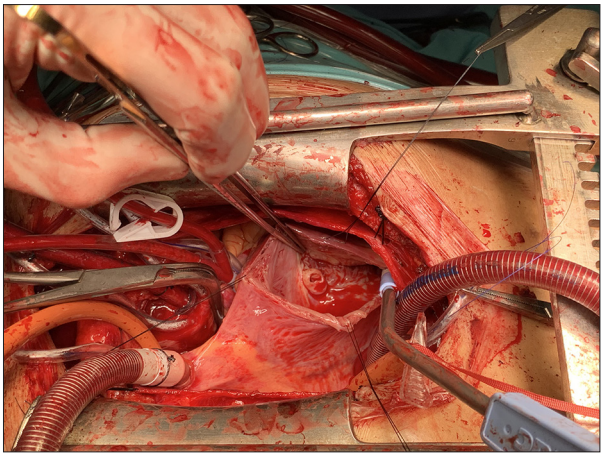

Figure 3. Type 2 Direct Gerbode defect. A direct Gerbode type VSD was observed on the membranous septum from the right atrium. No involvement of the defect was observed on the tricuspid valve (blue dashed line). The defect had a diverticulated appearance.

Surgical Techniques and Findings

The author performed surgical repair with median sternotomy using cardiopulmonary bypass, aortic cross-clamp, and cold blood cardioplegia. Upon opening of the RA, a 10-mm septal defect was observed in the AV membranous septum adjacent to the trigonum of Koch area and the annulus of tricuspid septal leaflet (**Figure 3**). The lesion appeared diverticulated with an opening of 1 cm in diameter and prominent blood flow gushing from the LV to RA. The saline test of the TV revealed mild regurgitation with a flail observed on the edge of the tricuspid septal leaflet. The Gerbode defect was closed using polytetrafluoroethylene (PTFE) patch with 9 circular pledget sutures (**Figures 4, 5**). The TV was repaired using De Vega technique.

Outcome and Follow-up

The patient was transferred to the ICU after surgery. There was no post-operative morbidity such as stroke, dysrhythmia, and acute kidney injury. The patient stayed in the ICU for 1 day and then was transferred to the intermediate ward for further

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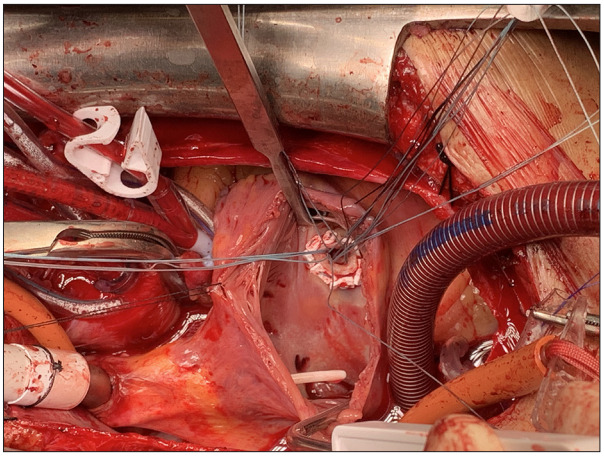

Figure 4. Surgical closure of the Gerbode defect. The Gerbode type ventricular septal defect was closed with 9 circular pledget sutures.

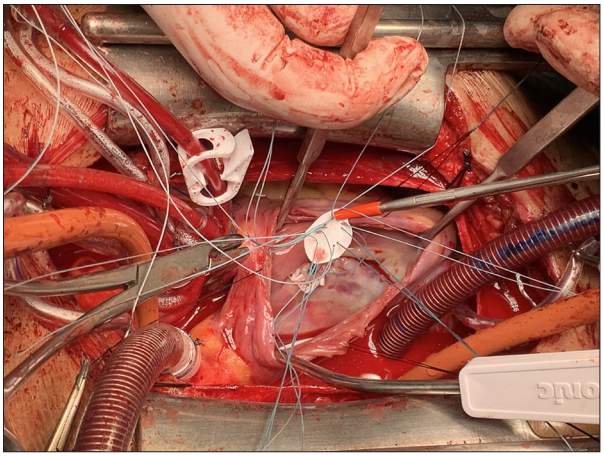

Figure 5. Surgical closure of the Gerbode defect. Defect closure with polytetrafluoroethylene (PTFE) patch.

care. Follow-up transthoracic echocardiography 4 days after surgery showed no residual VSD flow, a trace of residual tricuspid regurgitation, and global wall motion normokinetics.

Discussion

Gerbode defect is a rare type of VSD which was historically identified as a congenital heart anomaly. However, it is becoming more common as an acquired defect which can be either iatrogenic or non-iatrogenic. In a patient with no significant previous history of cardiac disease, the finding of a Gerbode defect suggests the congenital type lesion. The high pressure in the LV results in the shunting of blood from the LV to RA in Gerbode defects due to the large pressure gradients. The end results of dilated RA can be misinterpreted as tricuspid regurgitation during echocardiographic examination, which warrants further careful assessment of this condition to be able to totally exclude a Gerbode defect.

Patients with a Gerbode defect usually present various signs, but the most common are heart murmur and RA dilation [3]. The criterion standard to diagnose Gerbode defect is transthoracic echocardiography with color Doppler to visualized the shunt flow, but it can be misdiagnosed as tricuspid regurgitation [6]. Small asymptomatic defects may be treated conservatively but larger defects require surgical or interventional treatment [3]. Most common surgical intervention is patch repair, but there are other authors who prefer to use a pledgeted suture or direct suture [3,7].

The patient presented in this case report had no history of heart disease, yet his medical history was significant only for essential hypertension. The patient recently experienced shortness of breath and fatigue, which were getting worse during the last 2 years. Further evaluation confirmed the diagnosis of a direct type Gerbode defect (Type 2). Small Gerbode defects may be asymptomatic, and larger defects are usually associated with heart failure, but not in the present patient [3,8]. However, it is unclear whether the heart failure was caused by the Gerbode defect or by his chronic hypertension. The echocardiography showed a concentric left ventricular hypertrophy, which is a sign of an increased myocardial workload, which might be caused by his chronic hypertension. Furthermore, VSD is usually associated with eccentric hypertrophy, not concentric hypertrophy of the left ventricle; therefore, the clinical signs and symptoms in this patient were unlikely to have been caused by the Gerbode defect [9].

The incidental finding of the congenital type of the Gerbode defect was once reported in a diabetic patient who presented with additional symptoms of shortness of breath and fatigue [10]. As the left-to-right shunt from this lesion may complicate into pulmonary hypertension and potentially heart failure, thorough cardiac evaluations must be made in evaluating patients with signs and symptoms related to these abnormalities and with no prior history of cardiac diseases. The prompt identification of a Gerbode defect warrants a proper intervention with surgical repair, which may prevent further complications. This case report supports the importance of further studies to develop effective diagnostic screening methods to identify congenital heart diseases.

During surgery, the Gerbode defect that connected the LV to the RA showed an unusual pattern of VSD in which the lesion appeared to be diverticulated towards the RA. No other significant findings suggested that the lesion was an acquired defect, such as previous cardiac surgery or signs of infective endocarditis. Therefore, it is most likely that the defect was congenital in origin. However, the outpouching feature of the defect is an unusual finding, which was assumed to be due to the chronic high pressure of blood flow from the LV. There are currently no reported cases of a Gerbode defect with a similar feature. There are several possibilities of how the defect

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could appear diverticulated. First, the defect might have been very small initially and may not have caused any significant signs and symptoms. The chronic exposure of blood flow from the LV then led to the enlargement of the defect while at the same time pushed the tissue surrounding the lesion towards the RA. This mechanism would have made the early diagnosis challenging before the patient eventually developed complications from the defect, such as congestive heart failure.

Another possible explanation is that the patient might have had a congenital abnormality involving the membranous AV septum as a thinner or weakened membrane in a specific region with no communicable defect. However, the high-pressure gradient between the LV and RA may have over time pushed the weakened area of the AV septum, leading to diverticulum formation and introducing a defect connecting the LV to the RA. This phenomenon might also occur in the case of congenital cardiac diverticulum. Cardiac diverticulum in the left ventricle itself is a very rare congenital heart disease, with only several cases reported [11,12]. The diverticulated feature of the Gerbode defect in this patient raises the possibility of cardiac diverticulum as the underlying pathogenesis instead of the VSD.

Furthermore, although the rare Gerbode defect identified in our patient was Type 2 (direct), which does not involve the tricuspid leaflet, the defect seemed to affect the integrity of the tricuspid valve as indicated by the flail tricuspid leaflet with mild regurgitation. The condition was successfully repaired during the surgery. This finding also emphasizes the need for thorough evaluation of the overall cardiac function in the Gerbode defect during the reparation surgical procedure.

Conclusions

Congenital heart disease is currently one of the most common congenital diseases that require surgical intervention. The vast spectrum of severity in many congenital heart diseases makes the diagnosis and early recognition challenging, so many patients live with the condition undiagnosed until adulthood when further complications have occurred. The patient presented in this report showed an unusual diverticulated appearance of the Type 2 Gerbode defect, which most likely developed congenitally from cardiac diverticulum and complicated into the Gerbode defect.

Acknowledgement

The author would like to thank and acknowledge the contribution of the surgical team and the Intensive Care Unit at National Cardiovascular Center Hospital, Jakarta, Indonesia for patient care during surgery and hospitalization.

Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

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