



## The Gerbode Defect a Rare Congenital Structural Heart Disease- A case report

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### **Abstract**

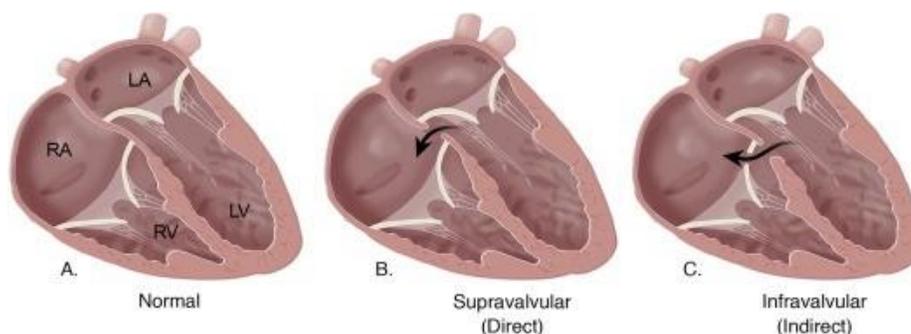
*Left ventricular (LV) to right atrial (RA) communication, also known as Gerbode defect, is very rare, usually congenital but sometimes also acquired. Cases of Gerbode defect have been reported after left valve surgery, usually valve replacement. We describe the first case of LV-RA communication not associated to valve surgery. The case we report concerns a 50-year-old woman for exertional shortness of breath. A LV-RA shunt was discovered echocardiographically. This case report confirms a congenital LV-RA shunt.*

**Keywords:** Gerbode Defect, Congenital Anomaly, Left to Right Shunt, Direct, Indirect, Ventricular Septal Defect, Tricuspid Valve

## Introduction:

A Gerbode defect is an acyanotic congenital heart disease with left ventricle to right atrium shunt. This defect was first reported by Meyer in 1857<sup>1</sup>. First successful closure was done by Kerby et al using hypothermia and inflow occlusion technique<sup>2</sup>. Dr. Gerbode described the first successful series of patients operated on with a Left Ventricle-Right Atrium shunt in 1958<sup>3</sup>. Riemenschneider and Moss have described two types of the communication based on the insertion of septal leaflet of Tricuspid valve, which divides the membranous septum into interventricular and atrioventricular parts<sup>4</sup>. According to STS congenital heart nomenclature<sup>5</sup> and database project the definition of Gerbode defect is a true Left Ventricle-Right Atrium communication.

There are two types, a direct and an indirect type as reported by Gerbode et al. The direct type of Gerbode defect is rare with a direct communication between Left Ventricle to Right Atrium<sup>4</sup>. It is sometimes referred to as supravalyvular defect (Figure 1). In Indirect type, there is a peri- membranous VSD and a defect in Tricuspid valve, the shunt is from left ventricle to right ventricle and then through the Tricuspid valve into Right atrium<sup>4</sup>. This communication occurs below the Tricuspid valve (Figure 1). In both defects, left ventricular outflow tract to right atrium communication allows for systolic shunting of blood to the right atrium. If this is a larger one, it can lead to volume overload and chamber enlargement<sup>6</sup>.



**Figure 1:** Different types of Gerbode defect.

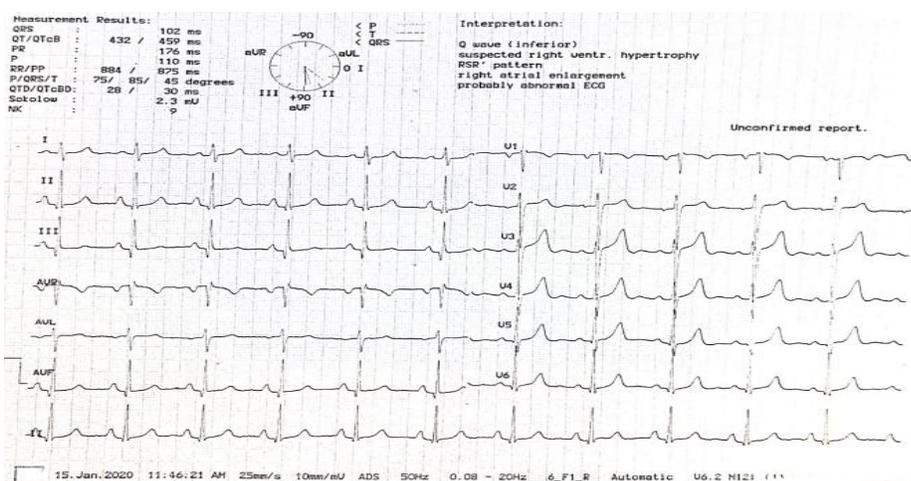
## Case Summery

A 50-year-old lady was referred from a primary health center who presented with exertional dyspnea, occasional left sided chest pain with radiation to arms & jaw, tingling sensation in legs and disturbed sleep.

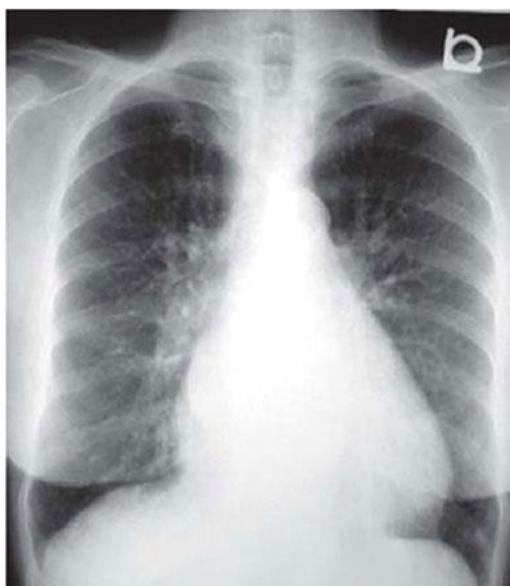
Clinical examination revealed an overweight middle- aged lady with a grade V/VI pansystolic murmur heard all over precordium best in 2<sup>nd</sup> to 4<sup>th</sup> intercostal space, with no signs of pulmonary hypertension or heart failure. An electrocardiogram revealed sinus rhythm with right atrium enlargement (Figure 2). Her chest x-ray showed mild cardiomegaly suggesting of right atrial enlargement (Figure 3). A

transthoracic echocardiogram revealed a non- dilated left ventricle, but a dilated right atrium and a 0.5 cm peri-membranous subaortic ventricular septal defect with left to right shunt, but the jet was directed into the right atrium. Close observation revealed septal tricuspid leaflet completely covering the ventricular septal defect by forming an aneurysm and directing a large turbulent jet into the right atrium through it (Figure 4).

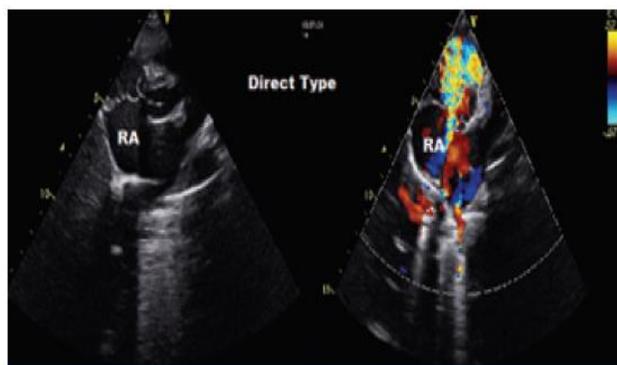
No vegetations were noted. There was neither a ruptured sinus of Valsalva nor an endocardial cushion defect seen. The continues wave Doppler of left ventricle–right atrial jet showed a gradient of 90 mmHg. Initially it was thought to be a tricuspid regurgitation due to pulmonary hypertension because there was grade-II tricuspid regurgitation and the calculated pulmonary artery systolic pressure was 50-60 mmHg. Patient refused a transesophageal echocardiographic examination and also any kind of surgery due to financial issues. She is maintaining well at 6-months follow up.



**Figure 2:** Electrocardiogram shows normal sinus rhythm with feature of right atrial enlargement.



**Figure 3:** Chest X-ray shows mild cardiomegaly with feature of right atrial enlargement.



#### Congenital Heart Disease

1. Gerbode defect (Communication between LV and RA) with Lt to Rt shunt. Peak gradient across the defect : 90 mmHg.
2. No regional wall motion abnormality noted.
3. Good LV systolic function with LVEF: 60-65%.
4. Tricuspid regurgitation Gr.II
5. Moderate pulmonary hypertension (PASP: 50-60 mmHg).
6. Normal RV systolic function.
7. Normal cardiac chamber in dimensions.
8. No evidence of LV apical thrombus / pericardial effusion.

**Figure 4:** Echocardiography shows presence of communication between LV and RA (Gerbode defect).

### **Discussion:**

The Gerbode defect is a very rare congenital anomaly. It is a rare defect representing less than 1% of congenital cardiac defects<sup>7</sup>. The defect is so rare that researchers observed only six cases of Gerbode Defect from 1990 to 2008 at Children's Memorial Hospital in Chicago<sup>8</sup>. This type of left ventricular-right atrial communication could result from a structural abnormality of the central fibrous body in combination with arrested maturation of the membranous ventricular septum<sup>9</sup>. The first description of a direct communication between the left ventricle and the right atrium was reported by Buhl in 1857<sup>1</sup>. The first successful closure of such a defect was reported by Kirby at the Hospital of the University of Pennsylvania in 1956<sup>2</sup>. The first successful series of patients operated on with a left ventricular-to-right atrial shunt was reported by a surgeon Frank Gerbode at Stanford University. There are two types known, a direct and an indirect as reported by Gerbode et al.<sup>3</sup>

#### **Type I (Indirect type)**

In a peri-membranous ventricular septal defect the shunt is from left ventricle to right ventricle then through the tricuspid valve into the right atrium. The communication thus occurs below the tricuspid valve. This is referred to as an indirect left ventricular to right atrial shunt.

#### **Type II (Direct type)**

In a true or direct Gerbode, the blood in the left ventricle goes through the small area of the membranous septum. This communication is above the tricuspid valve and left ventricle. It is rare than indirect. A true left ventricular-to-right atrial communication is the definition of a Gerbode defect according to the STS Congenital Heart Nomenclature and Database Project<sup>5</sup>.

We report one rare form of Gerbode defect that one having restrictive peri-membranous ventricular septal defect and left ventricle to right atrium jet through the small area of the membranous septum.

Due to apical displacement of tricuspid valve as compared to mitral valve, septal leaflet divides the membranous septum into interventricular and atrioventricular part; thus, the atrioventricular septum separates the left ventricle from the right atrium<sup>10</sup>. Another way of describing the defect is by using the classification into the supra-valvular left ventricle and infra-valvular left ventricle defects by Riemenschneider and Moss<sup>4</sup>. In this classification based on the anatomical relationship of the left ventricle to right atrium shunt with the tricuspid valve, the supra-valvular defects are in the atrio-ventricular septum while the infra-valvular defects occur between the left and right ventricles and then to the right atrium through a defect in the tricuspid valve. These valve defects can be due to leaflet perforations, malformation, widened commissure; or clefts.

The Gerbode defect is a ventriculo-atrial defect, the large systolic pressure gradient between the left ventricle and the right atrium is most likely the cause of the high velocity systolic flow from the left ventricle into the right atrium. A high Doppler gradient is one of the hallmarks of the Gerbode ventriculo-atrial defect because of the difference between the left ventricular systolic pressure and the low right atrial pressure.

In the direct variety, the defect is in the membranous part of the ventricular septum above the tricuspid valve, thus shunting the blood directly from the left ventricle to right atrium<sup>4</sup>.

The indirect Gerbode defect, is the commoner form of the defect. In this defect, the blood is shunted from left ventricle to right ventricle through a ventricular septal defect and from right ventricle to right atrium through defective tricuspid valve. Thus, the shunting of blood occurs indirectly from left ventricle to right atrium.

In both forms of defects, blood is shunted to right atrium during ventricular systole as there is significant pressure difference between left ventricle and right atrium during systole. This jet of blood in right atrium often mistaken as tricuspid regurgitation jet of pulmonary hypertension. This left to right shunt can lead to volume overload and chamber enlargement if it is large. This is unlike shunt from aorta to right atrium in ruptured sinus of Valsalva, which occurs both in systole and diastole. As aortic systolic and diastolic pressure are both significantly greater than that of right atrium.

The patient was relatively asymptomatic, however in a series<sup>8</sup> between the years 1990 and 2008 at Children's Memorial Hospital, all six patients (2 males and 4 females) who underwent closure of a direct congenital Gerbode-type ventriculo-atrial defect were symptomatic. In this series the size of the ventriculo-atrial defect ranged from 4 to 8 mm, with a mean size of  $6.2 \pm 2$  mm; while in our patient the Gerbode defect was 4 mm and the peri-membranous ventricular septal defect was 5 mm. The surgical outcome of this rare defect is excellent<sup>8</sup>.

In a review by Yuan SM, etiologies of the LV-RA shunts were congenital in 26.4% and acquired in 72.7% cases. Most of the acquired LV-RA shunts are of either a postoperative or an infective aetiology.

Transthoracic echocardiography showed a 62.2% accurate diagnosis, 13.4% inconclusive diagnosis, 9.8% missed diagnosis, and 14.5% misdiagnosis rate<sup>10</sup>. Our patient appeared to be of congenital aetiology, since no previous history of infection or surgery present. The case was diagnosed on transthoracic echocardiography by changing various views, using pulse wave, continuous wave and color Doppler. This along with right atrial chamber enlargement, normal pulmonary artery end diastolic pressure and an unusually high Doppler echocardiogram gradient compared to the ventricular septal defect with shunting only at ventricular level led us to the diagnosis in our case. These are the useful learning clues, especially for novice echocardiographers.

The transesophageal echocardiography or cardiac catheterization were more accurate in diagnosis of this rare defect than transthoracic echocardiography<sup>10</sup>. Although, the clinical course of left ventricular-right atrial communication is similar to that of ventricular septal defect, it may be differentiated from it by the earlier onset of congestive failure<sup>4</sup>.

### **Conclusion:**

A delayed diagnosis may inevitably lead to worsened condition of the patient. Therefore, when an unexplained turbulent flow is visualized in the right cardiac chamber, the possibility of left ventricle-right atrium shunt should be kept in mind. The correlation of findings on echocardiography with each other can prevent the misdiagnosis of this rare defect.

### **Reference**

1. Meyer H. "Ueberangeborene Engeoder Verschluss der Lungenarterienbahn. Archiv für Pathologische Anatomie und Physiologie und für Klinische Medicin". 1857; 12 (6): 497–538. [Google Scholar].
2. Kirby C, Johnson J, "Zinsser H. Successful Closure of a Left Ventricular-Right Atrial Shunt". Annals of Surgery. 1957; 145 (3): 392–94. [PMC free article] [PubMed] [Google Scholar].
3. Gerbode F, Hultgren H, Melrose D, Osborn J. "Syndrome of left ventricular-right atrial shunt successful surgical repair of defect in five cases, with observation of bradycardia on closure. Annals of Surgery". 1958; 148 (3): 433–46. [PMC free article] [PubMed] [Google Scholar].
4. Riemenschneider TA, Moss AJ. "Left ventricular-right atrial communication". Am J Cardiol 1967; 19: 710–8.
5. Jacobs J, Burke R, Quintessenza J, "Mavroudis C. Congenital Heart Surgery nomenclature and database project: ventricular septal defect". The Annals of Thoracic Surgery. 2000; 69 (3): 25–35. [PubMed] [Google Scholar].

6. O. M. Cheema, A. A. Patel, M. C. Su, and D. J. Shah, "Gerbode ventricular septal defect diagnosed at cardiac MR imaging: case report," *Radiology*, vol. 252, no. 1, pp. 50–52, 2009.
7. Wasserman S, Fann J, Atwood J, Burdon T, Fadel B. Acquired "Left Ventricular- Right Atrial Communication Gerbode-Type Defect". *Echocardiography*. 2002;19(1):67-72.
8. Kelle A, Young L, Kaushal S, Elise Duffy C, Anderson R, Backer C. "The Gerbode defect: the significance of a left ventricular to right atrial shunt". *Cardiology in the Young*. 2009;19(S2):96.
9. McKay R, Battistessa S, Wilkinson J, Wright J. "A communication from the left ventricle to the right atrium: a defect in the central fibrous body". *International Journal of Cardiology*. 1989;23(1):117-23.
10. Yuan S. "Expert review Left ventricular to right atrial shunt (Gerbode defect): congenital versus acquired". *pwki*. 2014; 3:185-94.