

Gerbode Defect—A Rare Defect of Atrioventricular Septum and Tricuspid Valve

ABHAY TIDAKE¹, PRANIL GANGURDE², AJAY MAHAJAN³

ABSTRACT

Left ventricular to right atrial communications (the Gerbode defect) are rare types of ventricular septal defect and present as direct or an indirect type. We hereby, report two cases, one direct and another indirect type. Cardiopulmonary bypass surgery was done and a successful suture closure of ventricular septal defect using pericardial patch was performed.

Keywords: Cardiomegaly, Pansystolic murmur, Ventricular septal defect

CASE REPORT

CASE 1

An eight-year-old asymptomatic boy was referred from a primary health centre for echocardiogram in view of murmur.

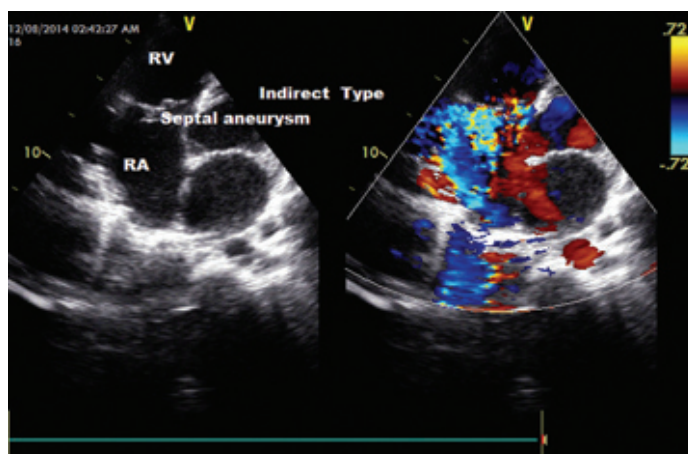
Clinical examination revealed an underweight child with a grade V/VI pansystolic murmur heard all over precordium best in 2,3,4th intercostal space, with no signs of pulmonary hypertension or heart failure. An electrocardiogram revealed sinus rhythm with right atrium enlargement. His chest x-ray showed mild cardiomegaly suggesting right atrial enlargement. A transthoracic echocardiogram revealed a non dilated left ventricle, but a dilated right atrium and a 0.4 cm perimembranous 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 [Table/Fig-1].

No vegetations were noted. There was neither a ruptured sinus of Valsalva nor an endocardial cushion defect seen. The continuous wave Doppler of left ventricle–right atrial jet showed a gradient of 69 mmHg. Initially it was thought to be a tricuspid regurgitation due to pulmonary hypertension. But there was trivial pulmonary regurgitation and the calculated pulmonary artery end diastolic pressure was 15 mmHg. Parents refused a transesophageal echocardiographic examination. Under cardiopulmonary bypass, he underwent successful suture closure of his ventricular septal defect using pericardial patch. The echocardiographic findings were confirmed intraoperatively. Septal tricuspid leaflet was found to form an aneurysm over the ventricular septal defect. There was perforation in septal tricuspid leaflet. He was doing well at 6-month follow up.

CASE 2

A 10-year-old male child born out of non-consanguineous marriage presented with chief complaints of progressive exertional breathlessness, fever, dry cough since 2 months.

On examination, apex beat was in 5th intercostal space ½ inch lateral to mid clavicular line, hyperdynamic, prominent systolic thrill, present all over the parasternal area especially in pulmonary, tricuspid, mitral areas and absent over carotids. On auscultation first and second heart sounds muffled in all areas, pansystolic murmur grade V/VI heard all over precordium best in 2,3,4th intercostal



[Table/Fig-1]: A transthoracic echocardiogram revealed a nondilated left ventricle, but a dilated RA and a perimembranous subaortic ventricular septal defect with left to right shunt, but the jet was directed into the RA. 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



[Table/Fig-2]: A transthoracic echocardiography of a perimembranous ventricular septal defect of size 5 mm with left to right shunt and an additional small to medium sized ventricular septal defect where the color Doppler jet travels from the left ventricle into the right atrium

space, widely split second heart sound. Chest radiogram suggestive of cardiomegaly with increased pulmonary blood flow. Transthoracic echo suggestive of a perimembranous ventricular septal defect of size 5 mm with left to right shunt and an additional small to medium sized ventricular septal defect–left ventricle to right atrium shunt [Table/Fig-2]. The continuous wave Doppler of left ventricle–right atrium jet showed a gradient of 74 mmHg. There was trivial pulmonary regurgitation and the calculated pulmonary artery end diastolic pressure was 14 mmHg. The echocardiographic findings were confirmed intraoperatively. Under cardiopulmonary bypass, he underwent successful suture closure of his ventricular septal defect using a pericardial patch. Patient was doing well on follow up.

DISCUSSION

The Gerbode defect is a very rare congenital anomaly. It is a rare defect representing less than 1% of congenital cardiac defects [1]. 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 [2]. 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 [3]. The first description of a direct communication between the left ventricle and the right atrium was reported by Buhl in 1857 [4]. The first successful closure of such a defect was reported by Kirby at the Hospital of the University of Pennsylvania in 1956 [5]. 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 2 types known, a direct and an indirect as reported by Gerbode et al., [6].

Type I (indirect type)

In a perimembranous 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 [7].

We report two rare forms of Gerbode defect. One in which a perimembranous ventricular septal defect is restricted by the adherent septal tricuspid leaflet at the anterocommissural area resulting in an left ventricle to right atrium jet through the tricuspid valve. Second one having restrictive perimembranous 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 [8]. Another way of describing the defect is by using the classification into the supralvalvular left ventricle and infravalvular left ventricle defects by Riemenschneider and Moss [9]. In this classification based on the anatomical relationship of the left ventricle to right atrium shunt with the tricuspid valve, the supralvalvular defects are in the atrioventricular septum while the infravalvular 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 Gerbode's defect, 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 [9].

The indirect Gerbode defect (case1), 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.

Both the patients were relatively asymptomatic, however in a series [2] 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 ± 2 mm; while in our patient the Gerbode defect was 4 mm and the perimembranous ventricular septal defect was 5 mm. The surgical outcome of this rare defect is excellent [2].

In a review by Yuan SM, aetiologies 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% inclusive diagnosis, 9.8% missed diagnosis, and 14.5% misdiagnosis rate [10]. Both of our patients appeared to be of congenital aetiology, since no previous history of infection or surgery present. In first case, it was misinterpreted as tricuspid regurgitant jet of pulmonary artery hypertension and second case was challenging due to the presence of additional shunt at ventricular level. Both the cases were diagnosed on transthoracic echocardiography by changing various views, using pulse wave, continuous wave and colour 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 lead us to the diagnosis in our cases. 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 [10]. 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 [9].

CONCLUSION

A delayed diagnosis may inevitably lead to worsened condition of the patient. Therefore, when an unexplained turbulent flow is visualised 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.

REFERENCES

- [1] 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.
- [2] 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.
- [3] 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.
- [4] Meyer H. Ueberangeborene Enge oder Verschluss der Lungenarterienbahn. *Archiv für Pathologische Anatomie und Physiologie und für Klinische Medizin*. 1857;12(6):497-538.

- [5] Kirby C, Johnson J, Zinsser H. Successful Closure of a Left Ventricular-Right Atrial Shunt. *Annals of Surgery*. 1957;145(3):392-94.
- [6] 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.
- [7] 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.
- [8] Silbiger J, Kamran M, Handwerker S, Kumar N, Marcali M. The Gerbode Defect: Left Ventricular to Right Atrial Communication-Anatomic, Hemodynamic, and Echocardiographic Features. *Echocardiography*. 2009;26(8):993-98.
- [9] Riemenschneider T, Moss A. Left ventricular to right atrial communication. *The American Journal of Cardiology*. 1967;19(5):710-18.
- [10] Yuan S. Expert review Left ventricular to right atrial shunt (Gerbode defect): congenital versus acquired. *pwki*. 2014;3:185-94.

PARTICULARS OF CONTRIBUTORS:

1. Senior Resident, Department of Cardiology, LTMMC and LTMGH, Sion, Mumbai, India.
2. Junior Resident, Department of Cardiology, LTMMC and LTMGH, Sion, Mumbai, India.
3. Professor, Department of Cardiology, LTMMC and LTMGH, Sion, Mumbai, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Abhay Tidake,
Senior Resident, Department of Cardiology, LTMMC and LTMGH, Sion, Mumbai-400022, India.
E-mail: abhaytidake@gmail.com.

Date of Submission: **Mar 31, 2015**Date of Peer Review: **Jun 10, 2015**Date of Acceptance: **Jul 15, 2015**Date of Publishing: **Sep 01, 2015****FINANCIAL OR OTHER COMPETING INTERESTS:** None.