A case of direct congenital Gerbode defect misdiagnosed as left to right ventricle shunt and literature review

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Abstract

The direct congenital Gerbode defect was a rare type of ventricular septal defect (VSD) causing a communication between the left ventricle (LV) and right atrium (RA). In this case, only LV-to-right ventricle (RV)shunt was found preoperatively. But in operation, the defect was located at the interventricular septum between the tricuspid (TV) and mitral valves (MV) where the shunt was from LV to right atrium (RA) and septal TV dysplasia was found. The shunt might be LV-to-RA and the blood flow into the RV through the defect of TV. This article discussed why the preoperative ultrasound misdiagnosed this type of shunt and reviewed the literature of Gerbode defect.

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Abstract:

The direct congenital Gerbode defect was a rare type of ventricular septal defect (VSD) causing a communication between the left ventricle (LV) and right atrium (RA). In this case, only LV-to-right ventricle (RV)shunt was found preoperatively. But in operation, the defect was located at the interventricular septum between the tricuspid (TV) and mitral valves (MV) where the shunt was from LV to right atrium (RA) and septal TV dysplasia was found. The shunt might be LV-to-RA and the blood flow into the RV through the defect of TV. This article discussed why the preoperative ultrasound misdiagnosed this type of shunt and reviewed the literature of Gerbode defect.

Keywords: Gerbode defect; left ventricular-to-right atrial shunt; ventricular septal defect

CASE PRESENTATION

A 2-year-old girl was referred to our hospital because of cardiac murmurs found in 1 month after birth. She had sweating,poor appetite and a history of thalassemia with her mother. There were no other manifestations except 2/6 rough systolic murmurs found between left parasternal 3-4 ribs. Preoperative echocardiogram showed a patent foramen ovale (PFO) and an 11.2 mm perimembranous ventricular septal defect (VSD) at inflow and trabecular with septal tricuspid valve (TV) tissue attachment to interventricular septum, causing a 3.1mm high-velocity systolic flow shunt (4 m/s) from left ventricle (LV) to the right ventricle (RV) and the flow persisted to the diastole (Figure 1-3 and Video S1-S3). The tricuspid regurgitation (TR) was mild and the velocity was not measured. The pulmonary artery was a little wider and the pressure was normal according to a mild pulmonary insufficiency (PI)(1.42m/s). But dilation of RV or other chambers were not found even if the defect was so large for her.

Intraoperatively, the location of the VSD was not completely in conformity with preoperative echocardiography results. The VSD was 10mm×8mm and was not found at RV but at right atrium (RA). It was located just above the septal leaflet of the TV but below the mitral valve (MV) where the shunt was supposed to flow from LV to RA not RV. The coronary sinus was just below the VSD. The dilation of the tricuspid annulus was not found. Anterior and posterior leaflets of TV were normal. The middle part of septal TV was short and the leaflet adhered to interventricular septum. Septal leaflet of TV was dysplastic (Figure 4-5). She underwent the closure of the VSD with an autologous pericardial patch and surgical suture of PFO using mild hypothermic cardiopulmonary bypass (CPB). As for the septal TV dysplasia, it did not cause a severe regurgitation or increase the RV load after closure of the VSD, so it was not repaired. Her postoperative course was uneventful. In the follow-up after 3 months, postoperative echocardiography revealed no residual shunt. The mild TR could still be seen but it was reduced.

DISCUSSION

This type of VSD was named after Gerbode[1] in 1958.Initially,the author illustrated that the lesion consisted of a VSD at high ventricular septum and a defect or dysplasia of the septal leaflet of TV. The VSD located just above the septal leaflets of the TV but below the MV which caused the LV-to-RA shunt. The defect or dysplasia of the septal leaflet of the tricuspid valve allowed the blood in LV shunt to RA. But gradually VSD causing a LV-to-RA shunt were all called Gerbode defect whether it accompany with the defect of the septal leaflet of the TV or not[2]. The cause of the defect can be congenital or acquired. Classical congenital Gerbode defect was so rare that researchers observed only six cases at the Children's Memorial Hospital in Chicago between 1990 and 2008[2]. The incidence of congenital LV–RA communications was 0.08% of all catheterized congenital defects[3,4], and 0.12% in autopsy material[5]. Acquired LV-to-RA communication was increasingly reported. It was often complication of endocarditis[6], myocardial infarction[7], blunt chest trauma[8] or cardiac surgery[9]. The estimated incidence of the acquired defect was 0.65%[10].

Riemenschneider and Moss[11] had described two types of LV-RA communication as direct or indirect based on whether the shunt was though the tricuspid valve defect. According to the location of the defect, Sakakibara and Konno[12] classified it into 3 types. Type 1, also called the direct type, was a supravavular defect occurring in the atrialventricular septum above TV which shunted blood from LV into RA directly. Type 2 was an infravavular defect occurring in the membranous, muscular intraventricular septum or endocardial cushion below the TV. It produced a communication between two ventricles but the blood shunted into RV diverted to RA through the defect of TV, so it was called the indirect type. Type 3 is an intermediate defect with both supravalvular and infravalvular components. The type 2 and 3 may have many variants to septal leaflet anomalies of the TV, including a cleft, widened commissural space, perforation abnormal chordae and other deformities. Type 1 was common in acquired cases but congenital type was very rare[2,13]. This case was a congenital type 1 defect.

Echocardiography was the most useful diagnosis method and was noninvasive and radiation-free. But this type of VSD was often misdiagnosed. Reviewing the preoperative ultrasound images according to the anato-

mical structure, a LV-to-RA communication might shunt into RV from dysplastic septal tricuspid valve and LV-to-RA-to-RV annuls might form. The abnormal blood flow existed in RA and RV almost at the same time. It was misdiagnosed as LV-to-RV shunt and the LV-to-RA shunt might be misled by the mild tricuspid regurgitation.

In ordinary VSD, we could usually find the blood of LV went directly into the RV during the systole but would not last until diastole and the pulmonary artery congestion and RV enlargement would usually happen before the RA enlargement. In this case, the LV-to-RA-to-RV shunt was confused with direct LV-to-RV shunt. The blood from LV shunted into RA through the defect but the pressure gradient was not reduced immediately then the blood flowed into the RV through the defect of TV. The pressure gradient of these two types of shunts might be similar and the blood flow at this location was complicated. By the way, the direct LV-to-RV shunt was more common. It was difficult to distinguish and easy to be misdiagnosed. The LV-to-RA shunt might persist to diastole and systolic in RA. The LV-to-RA-to-RV shunt might exist in systolic and diastolic but might be more obvious during systolic. But for simple LV-to-RV shunt, the LV-to-RV shunt could be seen during systolic and only TR could be seen in RA during systolic. In slow motion mode, the shunt flowing into RA before RV might be seen but because the heart beat so fast and the location of the defect was so near to the TV that it was easy to be misdiagnosed as VSD below TV in routine clinical visiting. The LV-to-RA shunt might be missed in apical view but it might be more obvious in other view. Multi-sectional assessment was critical.

And the LV-to-RA shunt was often disturbance with TR. Because the shunt was originated from LV, the pressure gradient between LV and RV was higher than the regurgitation from RV to RA. If it was regarded as TR, the high speed of regurgitation was inconsistent with low PI velocity and the abnormal severe pulmonary hypertension would be erroneously diagnosed. And the TR happened in systole. LV-to-RA shunt would persist to diastole. Silbiger[14]et al described clues suggesting the presence of a LV-to-RA shunt include (1) atypical jet direction, (2) persistent shunt flow into diastole, (3) the absence of ventricular septal flattening, (4) the absence of right ventricular hypertrophy, and (5) the presence of a normal diastolic pulmonary artery pressure estimated from the PI velocity. In this case, the TR velocity had not been measured, the shunt was misdiagnosed.

There were other useful techniques to make auxiliary diagnosis. Three-dimension (3D) real-time ultrasound, Cardiac magnetic resonance (CMR) and contrast-enhanced computed tomography (CT) were the adjuvant techniques to reveal further detailed anatomical and physiological information, but it was expensive and uneasy to obtain for children. Cardiac catheterization was invasive, so it was not necessary for diagnosis unless to evaluate the hemodynamics and pressure.

Some researchers recommended treatment of this defect according to the severity of symptoms associated with the size, location, magnitude of shunt, flow volume, development time magnitude of shunt. Asymptomatic, without circulatory overload or small defects with insignificant intracardiac shunt could be managed conservatively by following up rather than surgery[2]. But in our opinion, continuous shunt not only increased the load of the heart and the risk of infective endocarditis, but also the surgery was safe, so repairment was advocated no matter whether the symptoms were significant or not. The LV-to-RA communication was corrected by surgery traditionally and surgical closure had been demonstrated to be with excellent outcome and recommended for closure of all direct defects[2]. The percutaneous transcatheter closure techniques had been used mostly in acquired cases[13,15]or high-risk surgical candidates due to previous valve replacement, advanced age, anticoagulation, and multiple comorbidities[16]. In this case, the location of the defect was close to the valve, Koch's triangle, and coronary sinus (CS). Transcatheter closure might be unsuitable. And if the defect of TV did not cause severe regurgitation and increase heart load, it was unnecessary to repair it.

Conflict of interest : There are no conflicts of interest related to this case report for any of the authors.

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Figure 1: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In apical view of this patient. The white arrow showed the shunt. LA=left atrium, LV=left ventricle, RA=right atrium, RV=right ventricle.

Figure 2: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In parasternal long-axis view of this patient.The white arrow showed the shunt. LA=left atrium, LV=left ventricle, RV=right ventricle.

Figure 3: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In parasternal short-axis view of this patient. The white arrows showed the shunt. LA=left atrium, RA=right atrium, RV=right ventricle.

Figure 4: In operation, the VSD was 10mm×8mm was not found at RV but at RA. It was located just above the septal leaflets of the TV but below MV. Septal TV dysplasia was found. RA=right atrium, RV=right ventricle, TV=tricuspid valve, MV=mitral valve, VSD=ventricular septal defect, CS=coronary sinus.

Figure 5: In operation, the VSD was closed with a continuous technique using 6-0 prolene suture with an autologous pericardial patch.

SUPPORTING INFORMATION:

Additional supporting information may be found online in the Supporting Information section.

Video S1: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In apical view of this patient. The yellow arrow showed the shunt. LA=left atrium, LV=left ventricle, RA=right atrium, RV=right ventricle.

Video S2: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In parasternal long-axis view of this patient. The yellow arrow showed the shunt. LA=left atrium, LV=left ventricle, RV=right ventricle.

Video S3: Two-dimensional transthoracic echocardiography. Color Doppler examination in the same view. In parasternal short-axis view of this patient. The yellow arrows showed the shunt. LA=left atrium, RA=right atrium, RV=right ventricle.