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Case report

The Gerbode defect: the first case associated with a persistent left superior vena cava in the coronary sinus, described in a premature newborn

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Abstract

Background: The Gerbode defect is an extremely rare type of congenital cardiac anomaly characterized by direct communication between the left ventricle (LV) and right atrium (RA), leading to a left-to-right shunt. It can be either congenital or acquired, and its clinical manifestations can range from completely asymptomatic presentation to heart failure. Transthoracic echocardiography is the preferred technique for its diagnosis, although the gold standard is transesophageal ultrasound. Surgery is often elective, but conservative treatment is possible in asymptomatic cases.

Case presentation: A Caucasian female was born after 30 weeks of gestation, with a history of intrauterine growth restriction and suspected fetal cardiopathy. In response to her prematurity, she required non-invasive respiratory support and surfactant replacement. Echocardiography showed a persistent left superior vena cava in the coronary sinus, an atrial septal defect with a left-to-right shunt, and indirect signs of pulmonary hypertension. During her fourth day of life, the infant still showed respiratory distress and was difficult to wean from respiratory support. A second echocardiogram offered a hemodynamic explanation with the presence of a communication between the LV and RA, known as the Gerbode defect. The consequent pulmonary overflow required aggressive diuretic therapy for an extended period, which conferred clinical stability

despite occasional signs of respiratory fatigue and feeding difficulties. When the child was 2 months old, she underwent palliative cardiac surgery with a pulmonary artery bandage, which was followed by corrective surgical intervention at 8 months of corrected age.

Conclusions: To our knowledge, this is the first case of the Gerbode defect associated with a persistent left superior vena cava in the coronary sinus, described in a premature newborn. The complexity of the case is remarkable because several comorbidities were linked to the pulmonary immaturity of the patient.

Keywords

Congenital Gerbode defect, prematurity, pulmonary hypertension, respiratory distress, tricuspid insufficiency, echocardiography.

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Background

The Gerbode defect is an extremely rare, anomalous communication between the left ventricle (LV) and right atrium (RA) [1]. The first autoptic description of this defect by Thurnam dates back to 1838 [1, 2]. More than 100 years later, Kirby et al. successfully executed the first living closure surgery [3]. Within 1 year (1958), Gerbode et al. named this anomaly the "Gerbode defect," and described it as follows: "the lesion consists of a high ventricular septal defect associated with a defect of the septal leaflet of the tricuspid valve which allows left ventricular blood to enter the right atrium" [4].

Epidemiology

Although this anomaly constitutes only ~0.08% of intracardiac shunts [5], it has a significant clinical and prognostic impact on the patient. The Gerbode defect is predominantly caused by a defect in the endocardial cushion or by a failure in the closure of the atrioventricular (AV) channel, and it is therefore also linked to the AV defect family. The latter represents only ~7% of all congenital heart diseases [1].

Classification

According to the *International Pediatric and Congenital Cardiac Code*, the Gerbode defect is classified as a ventricular septal defect (VSD) [6], although it is a defect at the level of the AV septum [7]. The current anatomical classification segregates the Gerbode defect into 3 types: type 1, a direct or supravalvular defect in the AV part of the membranous septum; type 2, an indirect or infravalvular defect in the interventricular part of the membranous septum; and type 3, an intermediate defect, in both the AV and the interventricular part [1]. The literature is unclear on the incidence of the 3 types; however, the majority of studies on this topic have recently shown approximate incidences of 76%, 16%, and 8% of the total cases, respectively [8, 9].

In a review published in 2014, Yuan differentiated the Gerbode defect into congenital (26.4%) and acquired defects (72.7%) (while defects of unknown origin accounted for 0.8% of the cases). The acquired defects are divided into post-infection and post-operative defects [10].

Embryology and pathophysiology

The septation of the 4 chambers of the heart occurs during the early stages of embryonic development. In particular, the interventricular septum of the formed heart is composed of a small membranous part and a much larger muscular part [11].

The Gerbode defect involves the AV portion of the interventricular septum, and causes abnormal communication between the RA and LV through the membranous part of that septum. The septal leaflet of the tricuspid valve also derives from the membranous part of the IV septum after a delamination process. This may explain the pathogenesis of the Gerbode type II and III defects. The tricuspid defects characteristic of the indirect defect sub-types can range from simple perforation to dysplasia or malformation, to widened commissures and clefts [1].

The process of septation is regulated by transcription factors, such as NKX2-5, GATA4, and TBX5 [12]. Several studies have reported correlations between genotypic variants of the genes encoding these proteins and hereditary septal defects [13]. A few studies have shown a correlation between the Gerbode defect and genetic variants [14], but further investigations are required to validate these results.

The primary pathogenic aspect of the Gerbode defect is its hemodynamic effect, in that it causes a high pressure gradient between the LV and RA, allowing significant passage of blood towards the RA. This leads to a large volumetric overload in the right sections of the heart, contrary to the effects of interventricular defects [15]. The main factors characterizing the shunt entity are the defect size and pulmonary vascular resistance. The increased right ventricular pre-load causes the overload of the pulmonary circulation and an increased volume in the left sections, with progressive dilatation of the LV.

Clinical manifestations

The Gerbode defect can have a wide clinical spectrum: from completely asymptomatic presentation to heart failure, which may compromise the patient's survival. This broad clinical spectrum mainly reflects the size and duration of the defect [1]. The commonest symptoms are dyspnea, respiratory distress, feeding difficulties, and failure to thrive. Dyspnea is caused by the overload of the pulmonary circulation, which is overwhelmed by the increased volume and pressure of blood [9]. Large defects cause symptoms related to right heart failure, such as weakness, fatigue, edema, and hepatomegaly [1].

The most common manifestations on physical examination are a systolic murmur (similar to that associated with VSD) that is pansystolic, grade III-IV, of harsh quality, and that radiates widely [9], jugular turgor, hepatomegaly, and edema, and these often indicate incipient right ventricle (RV) failure.

Instrumental evaluation

The diagnostic gold standard for the Gerbode defect is transesophageal echography [10]. However, transthoracic echocardiography is the most frequently used and most accessible technique, even though it is less sensitive. The most common echocardiographic findings are right atrial dilatation and high-speed flow (> 4 m/s), which originates at the membranous septum and is directed to the RA. The ventricular-atrial shunt is best visualized using an apical projection, with subcostal and parasternal short axis views [1]. The image acquired must be differentiated from other frequent pathological conditions, including endocardial cushion defects, VSD, and tricuspid regurgitation (TR) [16]. For instance, if the systolic shunt is misinterpreted as TR, a misdiagnosis of severe pulmonary arterial hypertension (PAH) is often made [1]. However, the detection of normal pulmonary artery pressure, estimated from the diastolic regurgitation on the pulmonary valve, allows us to exclude PAH and to recognize the Gerbode defect. Silbiger et al. [15] identified several key echocardiographic clues that are highly specific for the Gerbode defect. These include: (1) atypical jet direction; (2) persistent shunt flow into the diastolic phase; (3) lack of ventricular septal flattening; (4) no right ventricular hypertrophy; and (5) normal diastolic pulmonary arterial pressure (estimated from the pulmonic regurgitant velocity). An ultrasound evaluation of RA hypertension is another potentially important diagnostic and prognostic suggestion. Key diagnostic features include: (1) pulsating RA and left-shifted atrial septum; (2) inferior vena cava plethora; (3) systolic liver inversion of the venous flow; and (4) reversed diastolic flow from RA to LV [15]. Two-dimensional echocardiographic imaging usually detects none of the anatomical deficiencies of the tricuspid valve that accompany the infravalvular Gerbode defect. Nonetheless, an important clue to their presence is a high-frequency systolic fluttering of the tricuspid valve, which is easily detectable using M-mode echocardiography. Nowadays, 3-dimensional echocardiography allows greater anatomical characterization of tricuspid defects and provides excellent assistance in transcutaneous repair interventions [5, 17, 18].

Magnetic resonance imaging is a very useful tool for the study of septal defects. It allows detailed anatomical information to be obtained and can quantify the degree of shunt with great precision [1]. However, it may be challenging to perform because it is expensive, unavailable at some centers, and requires the pharmacological sedation of small children. Its use is also contraindicated for patients with non-contemporary pacemakers or implantable cardioverter defibrillators [5].

Although cardiac catheterization was used in the past to quantify the hemodynamic impact of septal defects, it has lost its diagnostic importance and is rarely used today in the decision-making process.

Treatment

Yacoub et al. suggested the surgical closure of all LV-RA defects to preclude the risk of consequent endocarditis [19]. The closure technique for the Gerbode defect consists of surgical intervention with patches, with or without the repair of the tricuspid flap. However, in asymptomatic cases where the defect is small, conservative treatment is often preferred [20].

Case presentation

We describe the case of a Caucasian female patient born by urgent caesarean section in response to maternal hypertension at 30 weeks of gestational age (GA). Two factors were noteworthy in the pregnancy history: the diagnosis of intrauterine growth restriction at 27 weeks of GA and the persistence of the left superior vena cava (PLSVC) connected to the coronary sinus, featuring RA dilatation and right ventricular predominance, highlighted at the fetal ultrasound at 28^{+1} weeks of GA. During the pregnancy, prophylaxis for respiratory distress syndrome (RDS) was given with betamethasone (full cycle). Soon after birth, the newborn required mask ventilation (FiO_2 max 50%, Apgar index 8-10), and was therefore hospitalized in the Neonatal Intensive Care Unit (NICU) with prematurity and the presence of respiratory distress, which required not-invasive ventilation support. Her auxological parameters were: weight 970 g (10^{th} percentile), length 33 cm (< 3^{rd} percentile), and head circumference 25.5 cm (10^{th}-25^{th} percentile). At 4 hours of life, the patient showed increased signs of respiratory distress associated with increased FiO_2 (> 30%), and bilateral hypodiaphania was observed on chest X-ray (**Fig. 1A**). Therefore, one dose of surfactant (200 mg/kg) was administered using the less-invasive surfactant administration (LISA) technique [21]. After this treatment, her FiO_2 level decreased (to 21%), reflecting an improvement in her respiratory dynamics. At this point, after initial stabilization, it was possible to perform echocardiography to verify the suspicion of heart disease posed in the prenatal period. The ultrasound examination confirmed PLSVC in the coronary sinus (**Fig. 2**) and showed an atrial septal defect/patent foramen ovale with a bidirectional shunt and indirect signs of pulmonary hypertension or, in detail, right ventricular dilation with a D-shaped interventricular septum and mild-to-moderate tricuspid insufficiency (TI) featuring high pressure in the RV (about 40 mmHg). Moreover, the patent ductus arteriosus (PDA) was 3 mm and was characterized by a bidirectional shunt and pulmonary hypertension Doppler pattern. Lung ultrasound showed signs of RDS and pulmonary immaturity caused by surfactant deficiency, but respiratory assistance with nasal continuous positive airway pressure (nCPAP) led to progressive improvement in the child's respiratory dynamics and good hemodynamic compensation.

On the fourth day of life, a systolic murmur of moderate intensity (2/6 Levine) with a high pitch was auscultated in all heart areas. During the

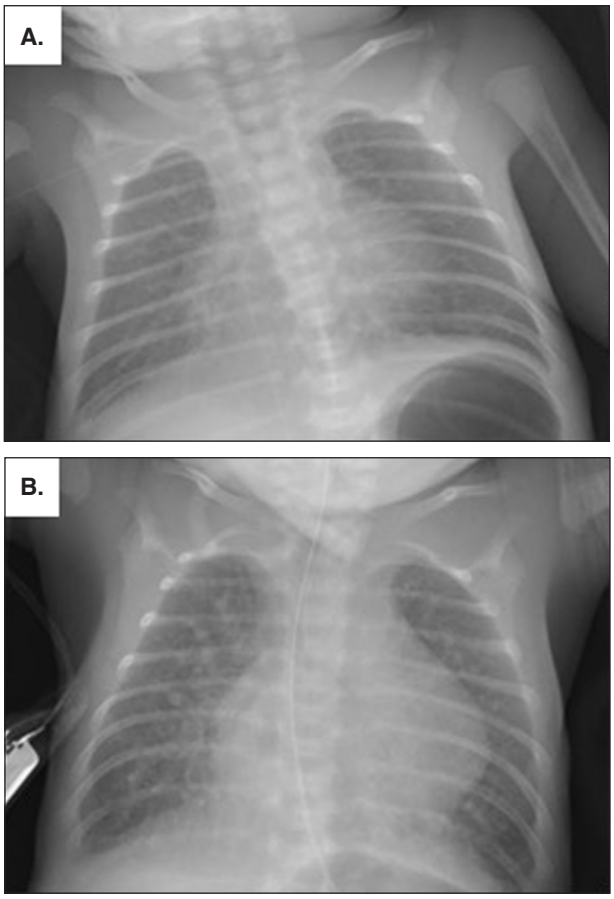

Figure 1. Chest X-ray. **A.** X-ray on the first day of life showing bilateral hypodiaphania. **B.** X-ray on the fourth day of life showing improvement in the pulmonary vascular weave and disappearance of the air bronchogram and cardiomegaly.

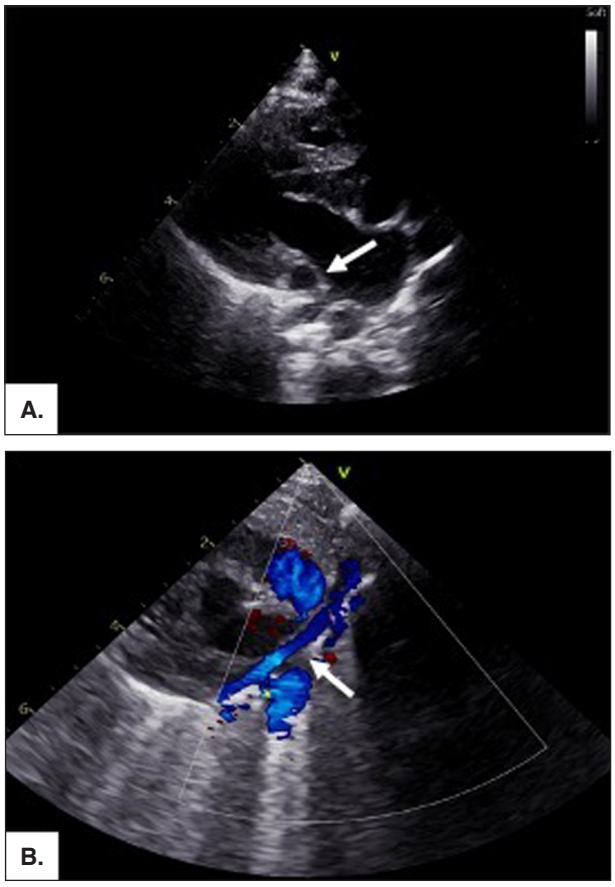

Figure 2. Echocardiographic image of the persistence of the left superior vena cava (PLSVC). **A.** Parasternal long-axis view showing classic dilatation of the coronary sinus (white arrow). **B.** Modified parasternal short-axis view showing PLSVC, which drains into the coronary sinus (white arrow).

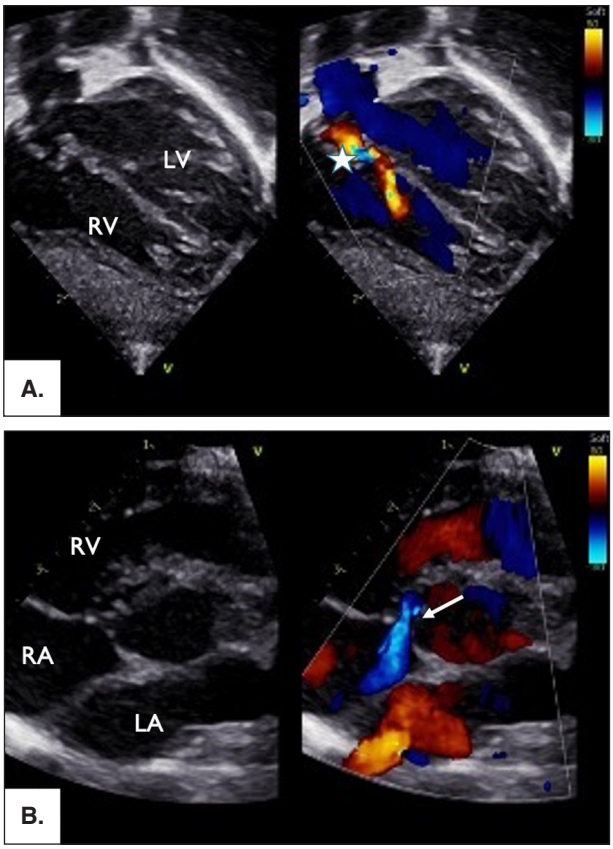

Figure 3. Double echocardiographic view of the same interventricular communication. **A.** Sub-costal long-axis view showing the interventricular defect with left-to-right shunt (red Doppler flow). **B.** Parasternal short-axis view showing the same defect but with the passage of blood into the right atrium (RA) in the systolic phase (blue Doppler flow).
LV: left ventricle; RV: right ventricle; LA: left atrium; RA: right atrium; star: interventricular communication; arrow: LV-RA communication.

following 24 hours, the infant showed worsening respiratory dynamics, although her hemodynamics were still good, evident as valid diuresis and no signs of hepatomegaly. A second X-ray showed improvement in the pulmonary vascular weave and the disappearance of the air bronchogram (**Fig. 1B**). An echocardiogram at this time showed initial left atrial dilation and the presence of a peri-membranous VSD, with a maximum gradient of 25 mmHg; initial right atrial dilatation with a systolic pulmonary artery pressure of 70 mmHg (estimated from TI), and a systemic blood pressure of 75/45 mmHg; the PDA was closed. Specifically, echocardiography showed a trans-tricuspid gradient through the VSD that was greater than the estimated pressure in the RV (about 50 mmHg). These findings, associated with right atrial dilation, which is more typical of pre-tricuspid defects, can be explained hemodynamically only by a communication between LV and RA; i.e., the Gerbode defect (**Fig. 3**). Therefore, diuretic therapy with low-dose furosemide (0.5 mg/kg, twice daily) was commenced, according to the radiological and ultrasound evidence of pulmonary hyperflow, and in view of the infant's actual clinical hemodynamic stability.

At 15 days of age, the infant still showed reduced lung compliance, indicated by the presence of polypnea, subcostal retractions, and an inability to be weaned from non-invasive respiratory support. She also showed fatigue when fed by bottle, which required the use of a nasogastric tube. The dose of diuretic (furosemide) was increased progressively to 3 mg/kg/day. Unfortunately, this change in diuretic therapy did not improve her signs of respiratory distress, so it was necessary to supplement with captopril at 0.18 mg/kg/day. Treatment with ethacrynic acid (2.5 mg/kg/day) was commenced when the infant was 1 month old to replace furosemide. During this period, attempts to suspend nCPAP resulted in respiratory acidosis and a slight contraction of diuresis.

Based on the complexity of the case, further investigations for other possible dysmorphisms that could lead to a syndromic picture were conducted, but none was confirmed. A blood sample was collected for genetic testing and no aneuploidies, microdeletions, or microduplications were detected with microarray-based comparative genomic hybridization. The exome sequencing analysis shows a heterozygous mutation of the trafficking protein particle complex 11 (*TRAPPC11*) gene, encoding a component of the multiprotein TRAPP complex, involved in membrane trafficking.

Diuretic therapy allowed the patient to maintain some clinical stability, although occasional signs of respiratory fatigue and feeding difficulties were observed up to 2 months of life. The infant was not subjected to definitive surgical correction because her weight was below the limit required for this intervention. Therefore, because she could not be weaned from respiratory assistance, she underwent pulmonary artery palliative bandaging, according to Trusler's rule. This treatment was performed in a dedicated Regional Cardiac Surgery Center. It is noteworthy that the child still required respiratory support (nCPAP FiO_2 21%) and reached a weight of 2,310 g at 38 weeks of corrected age. About a month after the operation, the patient was transferred again to our NICU. She underwent diuretic therapy with ethacrynic acid (3.5 mg/kg/day), spironolactone (2.3 mg/kg/day), and captopril (1.6 mg/kg/day). She also required non-invasive respiratory support through high-flow nasal cannula (FiO_2 max. 35%). From an echocardiograph taken after surgery, it appeared that the interventricular defect was partially obstructed by subvalvular tricuspid tissue; and the AV jet maintained a high gradient (70 mmHg, with a maximum gradient of 25-30 mmHg), and the pulmonary bandage was functionally adequate. During the remainder of the hospitalization period, the infant remained clinically stable, with a regular (even if sub-optimal) weight increase. Withdrawal of respiratory support was possible at 3 months of age. When discharged, the patient was 4 months old and 45 weeks of corrected age. She weighed 2,960 g (< 3rd percentile) and was bottle-fed with breast milk supplemented with formula milk.

In the following months, serial echocardiographic examinations showed good function of the pulmonary bandage (Gmax 80 mmHg, systemic blood pressure 95/65 mmHg), reduced signs of pulmonary circulation overload, and progressive reductions in both LV-RA flow and VSD size (Gmax 45 mmHg). It is noteworthy that the patient still required treatment with captopril, furosemide, and spironolactone.

At 8 months of corrected age, the child had reached 5,700 g in weight and underwent definitive corrective surgery, although her stature and weight were below the 3rd percentile.

During cardiac surgery, performed under cardioplegia, the VSD was closed using a bovine pericardium patch. Intraoperative confirmation of a tricuspid cleft allowed antero-septal tricuspid commissuroplasty to be performed. During the intervention, PLSVC was also confirmed. The patient had a normal post-operative course, with no complications. She was discharged after a few days with recommendations for continued clinical and cardiological follow-up.

Discussion and conclusions

The Gerbode defect is an extremely rare heart disease, accounting for only 0.08% of congenital heart diseases [5]. It is classified under different typologies according to the anatomical defect [1]. The real incidence of the various subtypes is not currently known, and the current bibliographical references are outdated [22, 23]. Furthermore, with increasing numbers of surgical interventions, the increasing cases of acquired Gerbode defect have further obfuscated the real incidence of the congenital forms [10]. The case described here is a form of congenital Gerbode defect found in a premature infant. It is an indirect type, and is therefore characterized by an interventricular defect associated with a deficit of the tricuspid valve, which causes the direct passage of blood from the LV (at high pressure) to the RA (at low pressure).

The final effect is an overload of both pressure and volume in the right chambers, and therefore an overload of the pulmonary circulation. In this case study, the newborn was also premature and therefore characterized by an immature pulmonary parenchymal substrate. Therefore, she could not tolerate the pulmonary hyperinflow, showed early clinical signs, and was difficult to wean from respiratory assistance.

The infant also had a second anatomical defect, PLSVC. No similar associations have been reported in the literature.

Echocardiography is the most accessible imaging technique, especially for newborns. In this situation, it must be performed by expert pediatric field operators because the Gerbode defect is often

confused for eccentric TI or other conditions. The most common mistake is to interpret the direct blood flow into the RA as an indirect sign of pulmonary hypertension. There are numerous echocardiographic clues that can help in the differential diagnosis of the Gerbode defect. The most important of these are the persistent shunt flow into diastolic phase; the lack of other indirect signs of pulmonary hypertension (i.e., the absence of ventricular septal flattening or a D-shaped septum, the discrepancy between the pulmonary pressure measurement calculated from TI and that measured with other methods, such as the interventricular defect or the pulmonary regurgitant velocity); and no right ventricular hypertrophy.

The case described here required a palliative pulmonary bandage, together with significant diuretic therapy. During the on-going follow-up, the interventricular defect decreased in size, although the tricuspid defect persisted, which caused a significant amount of blood to pass into the RA. Therefore, it took additional surgical intervention to completely correct the defect. This intervention allowed the anatomical site of the tricuspid defect to be identified.

The association between *TRAPPC11* gene and cardiac impairments has already been reported in the literature. In particular, recent studies described the association between heterozygous mutations of the *TRAPPC11* gene and right ventricular hypertrophy [24]. Homozygous or compound heterozygous variants of the *TRAPPC11* gene have also been associated with congenital muscular dystrophy with childhood onset [25]. To date, however, there are no conclusive studies on the association of heterozygous *TRAPPC11* variants with more complex cardiac malformations, such as of our case. The variant of *TRAPPC11* found in our neonate has not yet been reported in the literature, therefore its role as a cofactor cannot be excluded.

Conclusions

The Gerbode defect is an insidious cardiac anomaly, especially when diagnosed in a premature infant. An increasing number of post-surgical acquired forms of the defect are currently reported in the literature, whereas only few cases of the congenital forms are reported [23]. The rare incidence of this anomaly makes it hard to recognize. Moreover, the cardiology literature usually only links the Gerbode defect to the direct subtypes.

To our knowledge, this is the first case of the Gerbode defect associated with a persistent left superior vena cava in the coronary sinus, described in a premature newborn. Numerous co-morbidities linked to the GA of the child compounded the difficulty in managing the defect itself and made the decision-making process more complex. Echocardiography allowed us to make a diagnosis and guided us through the management of this patient.

This defect rarely resolves spontaneously and requires surgery in most cases. There is a paucity of data on this topic in the literature, and it remains poorly understood. More studies are required to clarify the etiology and the true incidence of this pathology.

Abbreviations

AV: atrioventricular
GA: gestational age
LISA: less-invasive surfactant administration
LV: left ventricle
nCPAP: nasal continuous positive airway pressure
NICU: Neonatal Intensive Care Unit
PAH: pulmonary arterial hypertension
PDA: patent ductus arteriosus
PLSVC: persistence of the left superior vena cava
RA: right atrium
RDS: respiratory distress syndrome
RV: right ventricle
TI: tricuspid insufficiency
TR: tricuspid regurgitation
TRAPPC11: trafficking protein particle complex 11 gene
VSD: ventricular septal defect

Ethics approval and consent to participate

The study was approved by the Pediatric Medical Ethical Review Board of the Tuscany Region.

Consent for publication

Written informed consent for publication was obtained from the patient's parents.

Declaration of interest

The Authors declare that they have no competing interests. This research received no specific grant from funding agencies in the public, commercial, or not-for-profit sectors.

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