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Case Report

Diagnostic approach and treatment of ventricular septal defect associated with PDA, coarctation of aorta, hypoplastic aortic arch and multiple valvular heart disease in a tertiary center: An infrequent association ☆

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ABSTRACT

Ventricular septal defect is the most common congenital heart disease in children and is associated with patent ductus arteriosus in 1%-7% of cases. The coexistence of both malformities with hypoplastic aortic arch and aortic coarctation is even rarer. We present the case of a 6-year-old girl referred to our hospital because of dyspnea on feeding, recurrent respiratory infections, poor weight gain, and a heart murmur. The image studies revealed a ventricular septal defect, patent ductus arteriosus, severe hypoplasia of the aortic arch with critical stenosis of the proximal portion, severe dilatation of the pulmonary artery and pulmonary, mitral, tricuspid, and aortic regurgitation. We will discuss the diagnostic approach and treatment in a tertiary reference center for patients with cardiovascular diseases.

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Introduction

Despite advances in diagnosis and surgical care, congenital cardiac malformations remain one of the leading causes of pediatric mortality [1,2]. A ventricular septal defect (VSD) is an important lesion that can present isolated or as a component of a complex congenital cardiac malformation [3]. Regarding the patent ductus arteriosus (PDA), the hemodynamic consequences are markedly variable and depend on its size and the cardiopulmonary status of the patient [4]. On the other hand, aortic arch hypoplasia can also present as an isolated abnormality or be associated with other congenital heart defects, especially coarctation of the aorta (CoA), and leads to severe cardiovascular complications if left untreated [5]. Due to the lack of evidence of these malformations coexisting and the importance of early diagnosis for proper management, we consider relevant to present this case in which the natural history of the disease led to severe symptoms in the patient and a complex surgical was required.

Case report

A 6-year-old girl was referred to a cardiac reference center due to dyspnea on feeding, recurrent respiratory tract infections, poor weight gain, and an unspecified heart murmur. The pregnancy and neonatal period were normal, and she had no relevant personal or family history. Physical examination revealed tachypnoea, hyperdynamic precordium, a grade III/VI systolic murmur in the left sternal border, a reinforced S2, and diminished pulses in lower limbs.

A 12-lead electrocardiogram suggested right ventricular hypertrophy, showing right axis deviation, high R waves in the right precordial leads (V1 and V2) and S waves in the left precordial leads (V5 and V6). A transthoracic echocardiography (TTE) was ordered, which revealed a VSD of 26 × 20 mm (Fig. 1A), PDA (Fig. 1B), severe hypoplasia of the aortic arch (5 mm) with critical stenosis of the proximal and distal portion, severe dilatation of the pulmonary artery annulus and trunk, severe cardiomegaly and left ventricular ejec-

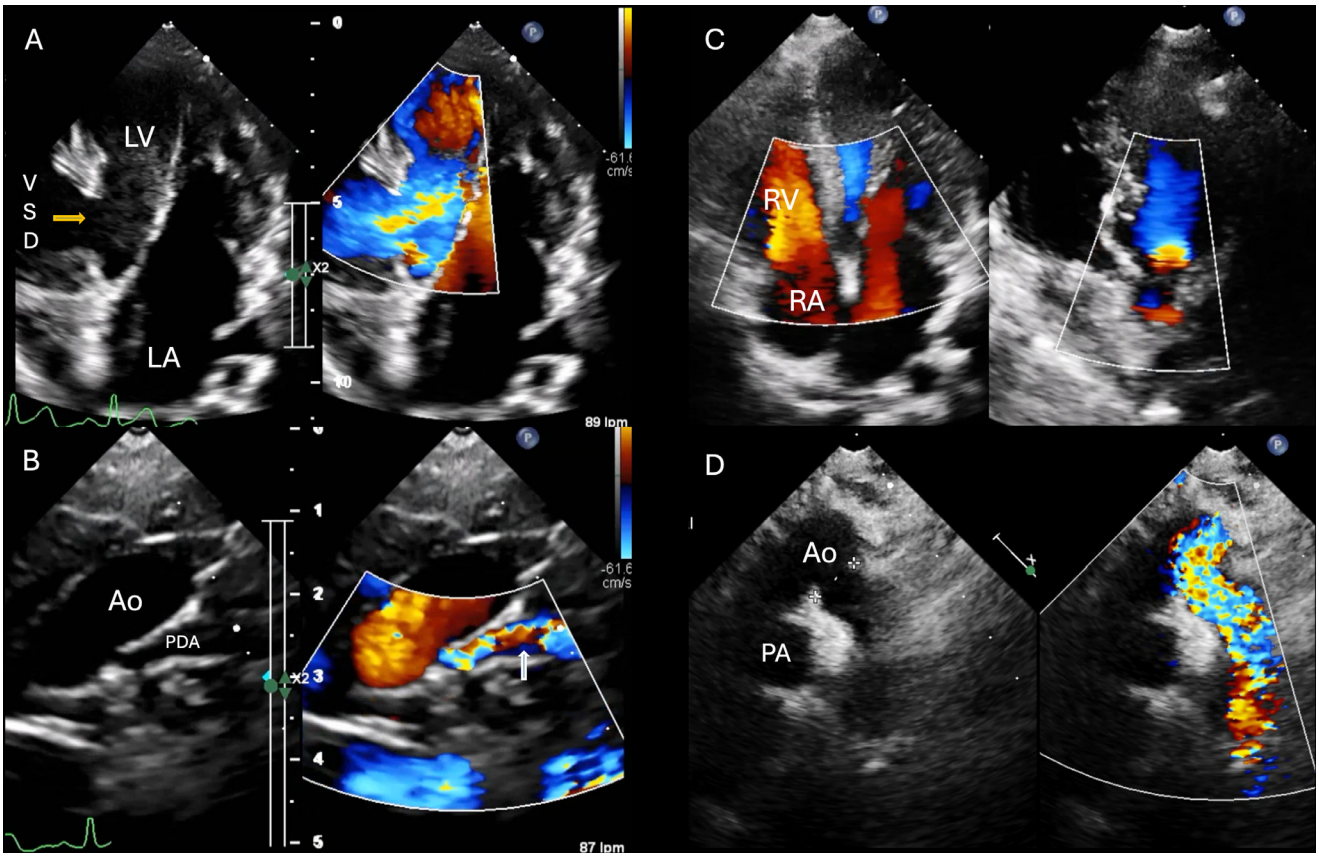

Fig. 1 – Pre- and postsurgical images by TTE 2D and color Doppler. (A) Pre-surgical 2D-4 chamber view showing a trabecular ventricular septal defect (yellow arrow) and left to right shunt. (B) Pre-surgical 2D-suprasternal view showing PDA (white arrow). (C) Postsurgical TTE 4 and 3-chamber views without ventricular residual shunt. (D) Suprasternal view of the aortic arch with PDA ligation. Abbreviations: LA, Left Atrium; LV, Left Ventricle; RA, Right Atrium; RV, Right Ventricle; Ao, Aorta; PA, Pulmonary Artery; PDA, Persistent Ductus Arteriosus; VSD, Ventricular Septal Defect.

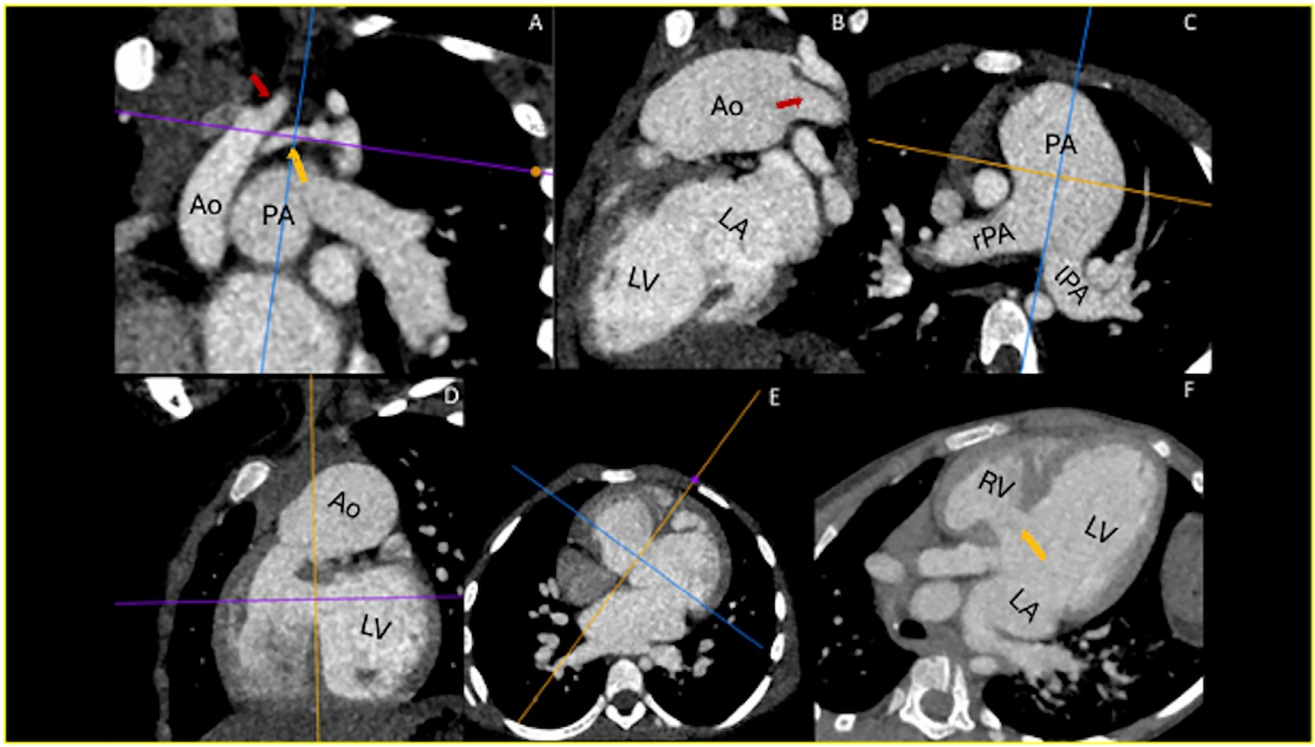

Fig. 2 – Computed tomography angiography revealing aortic arch hypoplasia (red arrow) of 5 mm corresponding to a Z score −6.79 and patent ductus arteriosus (yellow arrow). Aorta with stenosis at the aortic arch (red arrow) of 1.2 mm (B), dilation of pulmonary artery trunk of 35.5 mm with a Z score 5.15 (C), a ventricular septal defect (yellow arrow) of 26 × 20 mm (D, E) and general cardiomegaly (F). Abbreviations as before.

tion fraction (LVEF) of 61%. Computed tomography angiography confirmed the previous findings and revealed moderate pulmonary valve insufficiency and mild mitral, tricuspid, and aortic insufficiency (Fig. 2).

A multidisciplinary team was summoned to determine the management. The patient underwent aortoplasty with stent placement in the aortic arch and coarctation zone via therapeutic catheterization before undergoing surgery for VSD closure, aortic arch repair, and PDA ligation. After the surgery, the patient developed systemic hypertension, a possible complication of aortic arch hypoplasia surgical repair.

At the 3-month follow-up, the patient persisted with a systolic murmur in the left sternal border. However, follow-up TTE confirmed the absence of ventricular shunt (Fig. 1C) and a successful PDA ligation (Fig. 1D). Also, a gradient in the ascending aorta of 11 mm Hg and 10 mm Hg in the descending aorta, an unchanged LVEF (63%) and a pulmonary artery systolic pressure of 31 mmHg were reported.

Discussion

According to Yingjuan Liu and collaborators, between 1970 and 2017, the prevalence of cardiac heart defects globally increased by 10% every 5 years, with over 90% of this increase probably due to increased detection of lesions like VSD and PDA [6]. Moreover, this rise has also been accompanied by an increase of association of several cardiac malformations. While some simpler associations have been more commonly described (for example, the presence of both VSD and PDA or aortic hypoplasia with coarctation), several complex and multiple congenital heart diseases remain poorly assessed [7]. In this case we reported a patient that presented with the coexistence of a VSD, PDA, severe hypoplasia of the aortic arch and critical coarctation of the proximal portion.

Imaging assessments are indispensable for the accurate diagnosis and management of congenital heart defects such as VSD, patent ductus arteriosus PDA, and CoA. Echocardiography is the primary imaging modality used initially for all three conditions due to its noninvasive nature and capacity to provide detailed real-time images [8]. For VSD, echocardiography accurately delineates the location, size, and number of defects within the ventricular septum and assesses the shunt severity by visualizing the flow across the defect using color Doppler, which is essential for determining the hemodynamic impact [8,9]. PDA is identified by echocardiography through the visualization of the ductus arteriosus and the assessment of the aortic to pulmonary arterial flow, with Doppler imaging quantifying the flow velocity and direction to evaluate the physiological impact of the ductus [8,9]. In the assessment of CoA, echocardiography focuses on identifying the site of narrowing in the aorta and assessing the flow pattern across this re-

gion. It often shows a significant gradient across the coarctation site and may display left ventricular hypertrophy as a response to increased afterload. Additionally, the presence of collateral circulation may be suggested by abnormal flow patterns in intercostal or internal thoracic arteries. In fact, collateral circulation may be seen in a chest X-ray with the classical "Figure 3 sign", indicative of the indentation at the site of coarctation and post-stenotic dilatation of the descending aorta [10].

These findings are supported by further imaging with Cardiac MRI and CT angiography (CTA), which are utilized in complex cases or when detailed anatomical information is necessary for intervention planning [11]. CTA offers exceptional resolution for visualizing the aortic arch, detailing the extent of narrowing in CoA and revealing any associated anatomical features such as collateral vessels, which are crucial for comprehensive surgical planning. Cardiac MRI provides additional functional and structural details, including ventricular volumes and mass, beneficial in managing VSD and assessing vascular structures in PDA and CoA, offering a complete view of the cardiovascular system without ionizing radiation, thus suitable for repeated examinations in pediatric cases [11,12].

In the case of VSD, the increased right ventricular stroke volume will produce dilatation of the pulmonary artery and its branches, as well as increased pulmonary perfusion throughout the lungs [3]. If left untreated, the shunt formation can lead to hemodynamic compromise, pulmonary hypertension and even heart failure. Moreover, VSD with malalignment of the muscular outlet septum can cause a small part of the aortic root to override the crest of the muscular ventricular septum, which may result in aortic regurgitation or aortic valve prolapse [13,14].

In addition, the severity of aortic arch hypoplasia and the association with coarctation of aorta can be relevant to long-term risk of hypertension development [15]. Conversely, PDA can result in blood flowing from the descending aorta across the PDA into the pulmonary circulation and may result in pulmonary edema [16].

In conclusion, while there is no known direct relationship between VSD, PDA and aortic arch hypoplasia with coarctation of the aorta, these conditions can sometimes coexist leading to more complex congenital heart disease. The hemodynamic consequence of each malformation needs to be taken into consideration, especially in considering the management approach.

Patient consent

The authors confirm that written patient consent for publication has been obtained, in line with the COPE best practice guidelines, and that the individuals being reported on are aware of the possible consequences of the reporting. This study complies with the Declaration of Helsinki, and it was approved by the local ethics committee.

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