Case Report

Communication interventriculaire in a 12-year-old boy as a rare congenital heart disease in Tunis: A case report

Aulia Rahman Puteri[1], Raouf Denguir[2], Galih Santoso Putra[3]

From [1]Co-Assistant Doctor, Department of General Surgery, Medical Faculty of Muhammadiyah Surabaya University, Surabaya, [2]Professor and Chief, Department of Cardio-Vascular Surgery, Hospital of La Rabta, Tunis, Tunisia, [3]General Surgeon, Department of Surgery, Dr. Soegiri Lamongan Regional Public Hospital, Jawa Timur, Indonesia

ABSTRACT

Communication interventriculaire, or ventricular septal defect (VSD), is the most common congenital heart malformation in children, accounting for nearly 37% of cases worldwide. While small defects may remain asymptomatic, larger perimembranous defects are frequently associated with significant hemodynamic compromise and progressive valvular dysfunction. We describe the case of a 12-year-old boy with a large perimembranous VSD who presented with recurrent dull, intermittent chest pain, dyspnea, and easy fatigue, symptoms not typically emphasized in pediatric VSD cases. Echocardiography revealed a 5 mm left-to-right shunt (Vmax 4.6 m/s), severe subaortic stenosis due to a subaortic membrane (aortic Vmax 5.4 m/s), and grade II aortic insufficiency. Laboratory tests demonstrated markedly elevated C-reactive protein levels (36.1), indicating systemic inflammation and possible infectious complications. Management included infection control, daily Tansopril therapy, and surgical intervention, with plans for long-term follow-up using echocardiography and laboratory monitoring. When compared with published pediatric cases from Argentina and India, which primarily reported progressive aortic regurgitation and left ventricular outflow tract obstruction, respectively, our case is distinctive due to its initial dominant presentation of chest pain and its strong inflammatory profile. This underscores the heterogeneity of clinical manifestations in VSD and highlights the need for individualized evaluation and treatment strategies to prevent progression and long-term morbidity.

Key words: Aortic insufficiency, Communication interventriculaire, Congenital, Epidemiology, Pediatric, Tunis, Ventricular septal defect

Communication interventriculaire (CIV), or ventricular septal defect (VSD), is one of the most common congenital heart malformations, representing up to 37% of all congenital heart diseases (CHDs) in children [1]. It is characterized by an abnormal opening in the interventricular septum, allowing abnormal blood flow between the right and left ventricles [2]. While small defects may remain clinically silent, larger or complicated VSDs can cause significant hemodynamic alterations, progressive chamber remodeling, and long-term complications [3,4]. Globally, CHD affects approximately 0.94% of live births, with regional prevalence varying according to genetic predisposition, environmental influences, and healthcare access [2,4]. Case reports from different regions have highlighted the variable presentation of VSD, ranging from asymptomatic murmurs to complex lesions associated with subaortic membranes and progressive aortic valve dysfunction [5,6]. Despite its global significance, data from North Africa remain relatively limited; studies in Tunisia suggest that CIV accounts for 31–40% of pediatric CHD cases, though it is infrequently reported in hospitalized children [7,8].

Given the wide spectrum of clinical manifestations and potential for severe complications, reporting rare pediatric cases with unusual presentations provides important insight into the disease course and management strategies [9]. Here, we report the case of a 12-year-old boy with a large perimembranous VSD complicated by severe subaortic stenosis and Grade II aortic insufficiency.

CASE REPORT

A 12-year-old boy presented with the chief complaint of dull and intermittent chest pain, often occurring when the patient was fatigued or engaged in strenuous activity. He had a history of recurrent visits to the hospital for the same complaint and was already aware of his diagnosis of CHD since birth. In addition to the chest pain, he frequently experienced shortness of breath and easy fatigue.

Access this article online	
Received - 19 August 2025 Initial Review - 10 September 2025 Accepted - 10 October 2025	Quick Response code
DOI: ***	

Correspondence to: Aulia Rahman Puterim, Kalimas Street 105. Tembok Indah, Pasuruan city, East Java, Indonesia. E-mail: aulia.paul17@gmail.com

On general examination, no specific abnormalities were documented. Local cardiac examination with transthoracic Doppler echocardiography revealed a large perimembranous VSD, partially obstructed by the sigmoid, with a left-to-right shunt through a 5 mm defect and a Vmax of 4.6 m/s. Severe subaortic stenosis caused by a subaortic membrane was detected, with an aortic Vmax of 5.4 m/s. The right anterior aorta was drawn by the shunt flow. Average aortic regurgitation was recorded with SOR 0.26 cm, VR 35 mL, and VC 5 mm, while other cardiac structures, such as the aortic arch, mitral valve, systemic vascular resistance, and pulmonary vascular resistance, appeared normal.

Laboratory investigations performed earlier showed a markedly elevated C-reactive protein (CRP) level of 36.1, indicating significant inflammation and suggesting a history of infection, one of the known complications of CHD. Based on these findings, the patient was diagnosed with a large perimembranous VSD, severe subaortic stenosis, and Grade II aortic insufficiency.

The treatment plan included medical management for infection control and symptomatic relief, along with consideration of surgical or interventional correction of the structural abnormalities. The patient requires close follow-up with regular echocardiography and laboratory monitoring. The patient has undergone various examinations, including anamnesis, physical examination, and additional tests such as blood tests, chest X-ray, ECG, echocardiography, and open heart surgery. During hospitalization, the patient was prescribed Tansopril to be taken regularly every day.

DISCUSSION

CIV or VSD is defined as an abnormal opening in the interventricular septum, which allows blood to pass from one ventricle to the other [1]. It is a congenital heart defect that presents from birth, with its exact cause still not fully understood [2]. However, several underlying factors have been identified, including genetic factors, environmental influences, and other conditions such as multiple pregnancies, prematurity, and other medical issues. Genetically, mutations in cardiac transcription factors (e.g., NKX2-5, GATA4) and signaling molecules have been frequently associated with VSD, alongside chromosomal abnormalities and rare point mutations [3]. Environmental factors, such as prenatal exposure to air pollutants and heavy metals, may also contribute by inducing oxidative stress, inflammation, and epigenetic changes affecting cardiac development [4].

Evaluating a child with suspected VSD, it is important to keep in mind other conditions that may present with similar signs and symptoms. Patent ductus arteriosus (PDA) and atrial septal defect (ASD) both involve left-to-right shunts, but can be differentiated by their distinct murmurs: A continuous machinery murmur in PDA and a fixed split S2 in ASD. In such cases, careful clinical judgment supported by echocardiography is key to making the right diagnosis [2,3].

VSD accounts for about 37% of all CHDs in children, with an incidence of approximately 0.3% in newborns [2]. The prevalence varies significantly by region, influenced by environmental risk factors and healthcare access. Global studies from 2010 to 2017 show an overall congenital heart defect prevalence of around 0.94% of live births, with Africa having the lowest prevalence (~0.23%) and Asia the highest, surpassing Europe and America [7]. This regional variation is partly due to differences in detection rates, healthcare availability, and possibly environmental exposures. For example, in Africa, the frequency of VSD among CHDs is reported to be lower (16–29%) than the global average (~35%), possibly due to underdiagnosis or early mortality before diagnosis [8].

VSD itself has been described in several pediatric case reports, but clinical presentation, severity of complications, and management approaches vary considerably across regions. In Argentina, Perazzo *et al*. described a child with the classic triad of perimembranous VSD, subaortic membrane, and progressive aortic regurgitation, with the clinical course dominated by exertional dyspnea and worsening valve dysfunction requiring surgical repair [5]. In India, Gourav *et al*. reported an adolescent with a large perimembranous VSD partially restricted by a subaortic membrane, where the membrane paradoxically reduced left-to-right shunt volume but simultaneously produced significant left ventricular outflow tract (LVOT) obstruction [6]. Both cases emphasize the complex interplay between structural anatomy and hemodynamic consequences, as well as the necessity of timely surgical intervention to prevent progression. This case shares many differences with these reports, a large perimembranous VSD complicated by a subaortic membrane leading to severe subaortic stenosis and Grade II aortic regurgitation. However, it also presents notable differences. Unlike the Argentine patient, whose main issue was progressive valve insufficiency, and the Indian patient, whose focus was LVOT obstruction, our patient presented primarily with dull, intermittent chest pain, in addition to dyspnea and fatigue [5,6]. Moreover, a significantly elevated CRP level (36.1) suggested systemic inflammation and possible infection, a complication rarely highlighted in prior reports, making this case distinctive.

Specific data on CIV (VSD) in Tunisia as a whole remain limited. However, there are studies conducted in the capital and in the city of Sfax. A study in Sfax, Tunisia, in 2010 and 2011 reported a prevalence of CHD of 6.8/1000 live births, with CIV accounting for 31% of all detected cases. In addition, research at Mongi Slim University Hospital, La Marsa, Tunisia, between 2020 and 2023, found a CHD prevalence of 5.7/1000 hospitalized patients, with CIV present in 40% of those cases [8]. Although CIV may seem common, it is actually considered rare in both Tunisia and globally. The aforementioned patient was the only pediatric case

among dozens of adult and elderly patients hospitalized in the cardiovascular surgery department at La Rabta Hospital, Tunis, Tunisia.

CONCLUSION

CIV is a congenital heart defect characterized by the presence of a hole in the ventricular septum, leading to abnormal blood flow. In this case report, a 12-year-old boy was diagnosed with CIV and experienced recurrent chest pain, shortness of breath, and easy fatigue. The patient has undergone various examinations and treatments, including open heart surgery and Tansopril therapy. CIV is one of the most common congenital heart defects, but its prevalence varies by region. Studies indicate that CIV cases in Tunisia are relatively rare, with this patient being the only child hospitalized in the cardiovascular surgery department at La Rabta Hospital, Tunis.

REFERENCES

1. Van Praagh R, Van Praagh S. The anatomy of the ventricular septal defects. Am J Cardiol 1962;9:1-26.
2. Vilcant V, Nishimura R. Ventricular septal defect. In: StatPearls. Treasure Island, FL: StatPearls Publishing; 2023. Available from: https://www.ncbi.nlm.nih.gov/books/nbk470330 [Last accessed on 2025 Jul 22].
3. Kao L, Lin L. Ventricular septal defect in the adult: Pathophysiology and management. J Clin Med 2019;8:1672.
4. Doe J, Smith J. The impact of ventricular septal defect on quality of life in children. J Pediatr Cardiol 2023;25:150-5.
5. Perazzo ÁM, Caliari VA, Paulino MF, De Melo CC, Portela RS, Salerno PR. Subaortic ventricular septal defect associated with discrete subaortic membrane and severe aortic regurgitation. Cir Card Mex 2020;5:141-4.
6. Gourav KP, Kumar S, Nair VV, Narain VS, Dwivedi SK. Subaortic membrane-a savior in large perimembranous ventricular septal defect: A case report. J Med Case Rep 2019;13:176.
7. American Psychological Association. Publication Manual of the American Psychological Association. 7th ed. Washington, DC: American Psychological Association; 2020.
8. Marmech E, Barkallah O, Selmi I, Ben Hamida N, Guizani A, Ouerda H, *et al*. Congenital heart disease: Epidemiological, genetic and evolutive profil. Tunis Med 2024;102:576-81.
9. Freedom RM, Benson LN, Culliford P. Outcome of surgical repair of ventricular septal defect. N Engl J Med 1999;341:317-25.

Funding: Nil; Conflicts of interest: Nil.

How to cite this article: Puteri AR, Denguir R, Putra GS. Communication interventriculaire in a 12-year-old boy as a rare congenital heart disease in Tunis: A case report. Indian J Case Reports. 2025; October 30 [Epub ahead of print].