Research Article

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Uncorrected Ventricular Septal Defect Complicated by Severe Pulmonary Arterial Hypertension, Pulmonary Embolism, and Nephrotic Syndrome in a Young Adult: A Case Report from the Borgou Departmental University Hospital, Benin (2025)

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Abstract

Introduction: Uncorrected ventricular septal defects can progress to severe complications in adulthood, including pulmonary arterial hypertension and Eisenmenger syndrome. In subSaharan Africa, late diagnosis remains common.

Case Presentation: A 31-year-old man was admitted for exertional dyspnea and lower limb edema. Physical examination revealed a continuous systolic-diastolic murmur and signs of right heart failure. Laboratory tests showed severe renal failure (creatinine: 101.96 mg/L), nephrotic syndrome (hypoalbuminemia: 15.03 g/L, massive proteinuria), and an inflammatory syndrome. Transthoracic echocardiography revealed a 10-12 mm perimembranous ventricular septal defect with severe pulmonary arterial hypertension, right ventricular dilatation and hypertrophy, and intracardiac thrombi. Management involved multidisciplinary medical care, with surgical intervention being contraindicated due to fixed pulmonary hypertension.

Conclusion: This case illustrates the adverse outcome of untreated ventricular septal defects and underscores the importance of early screening and correction of congenital heart diseases in subSaharan Africa.

Keywords

Ventricular Septal Defect; Pulmonary Arterial Hypertension; Pulmonary Embolism; Nephrotic Syndrome; Adult; Benin.

INTRODUCTION

Ventricular septal defects (VSDs) account for 20-30% of all congenital heart diseases [1]. While the majority of small VSDs close spontaneously during childhood, uncorrected moderate to large defects can progress to severe complications in adulthood [2].

Pulmonary arterial hypertension (PAH) is the most feared complication of untreated VSDs, resulting from chronic pulmonary overperfusion leading to irreversible vascular remodeling [3]. Eisenmenger syndrome, the ultimate stage of this progression, contraindicates surgical correction [4]. In sub-Saharan Africa, late diagnosis of congenital heart diseases remains common due to limited access to specialized care [5]. We report a case illustrating the complexity of managing a complicated VSD at an advanced stage in a Beninese context.

CASE PRESENTATION

A 31-year-old male, an IT professional, with no known history of heart disease, was admitted for NYHA stage II exertional dyspnea and lower limb edema evolving over three weeks. His medical history included a severe head injury seven months prior and a sedentary professional lifestyle.

Clinical examination revealed a deteriorated general condition (WHO stage III), stable hemodynamic parameters (BP: 103/81 mmHg, HR: 92 bpm, SpO2: 97%), a 5/6 systolic-diastolic heart murmur audible at all foci, and signs of right heart failure.

Laboratory tests demonstrated severe renal failure (creatinine: 101.96 mg/L), nephrotic syndrome (hypoalbuminemia: 15.03 g/L, massive proteinuria 4.02g/24h, hypoproteinemia 44.90g/L), an

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inflammatory syndrome (CRP: 96 mg/L), and hyperkalemia (5.80 mEq/L).

The electrocardiogram showed right ventricular hypertrophy and repolarization abnormalities (Figure 1). Transthoracic echocardiography revealed a 10-12 mm perimembranous VSD with severe PAH, right ventricular dilatation and hypertrophy, and right intracardiac thrombotic masses (Figure 2), suggestive of pulmonary embolism.

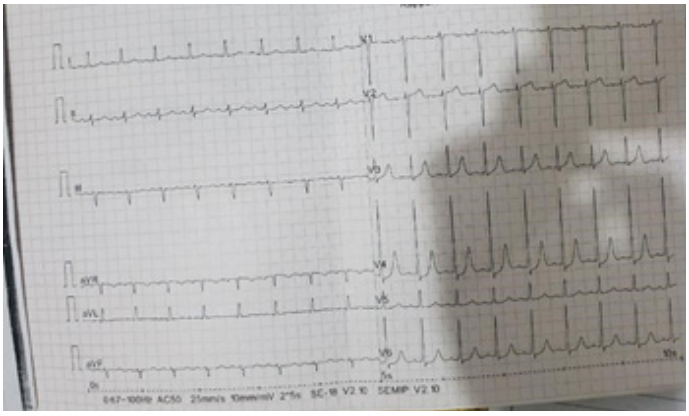

Figure 1: Electrocardiogram showing a regular accelerated sinus rhythm at 92 bpm, rightward deviation of the heart axis, right ventricular hypertrophy, and secondary extensive anterior repolarization abnormality.

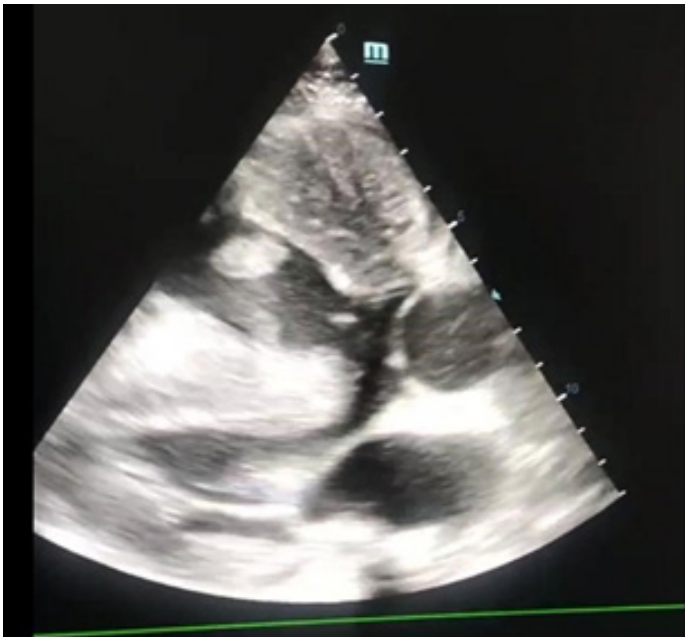

Figure 2: Image Echocardiography showing a perimembranous ventricular septal defect (10–12 mm), severe pulmonary hypertension, with right ventricular dilation and hypertrophy and the presence of right intraventricular thrombotic masses.

The final diagnosis was a long-standing congenital VSD complicated by severe PAH with chronic cor pulmonale, and pulmonary embolism in a prothrombotic state related to nephrotic syndrome. Initial management included diuretics, anticoagulation, correction of electrolyte imbalances, and multidisciplinary specialist consultations. Surgical evaluation was compromised by the severe, likely fixed, PAH.

DISCUSSION

Our observation eloquently illustrates the adverse natural history of uncorrected ventricular septal defects, with the development of severe pulmonary arterial hypertension and multisystem complications at a young age. This progression, well-described in the literature, remains particularly dramatic in Africa where therapeutic options are limited.

Pathophysiology of PAH in VSD: From Shunt to Fixed Hypertension

Pulmonary arterial hypertension associated with congenital heart disease results from a chronic left-to-right shunt leading successively to pulmonary overperfusion, reactive vasospasm, and irreversible vascular remodeling [6]. In our patient, the large VSD (10-12 mm) allowed a significant shunt for over three decades, leading to the establishment of severe, likely fixed, pulmonary arterial hypertension. Eisenmenger syndrome, the ultimate stage of this progression, is characterized by shunt reversal (right-to-left), explaining cyanosis and secondary polycythemia [4]. Although our patient did not present with frank cyanosis, the major right ventricular hypertrophy and dilatation observed on echocardiography attest to the chronicity and severity of the condition.

The association of VSD-PAH-pulmonary embolism represents a formidable complication with a multifactorial pathophysiology. In our patient, the prothrombotic state resulted from the combination of several mechanisms: blood stasis in the dilated and dysfunctional right ventricle, nephrotic syndrome with urinary loss of natural anticoagulant factors [7], and systemic inflammation evidenced by the significant elevation of CRP (96 mg/L). The presence of thrombotic masses in the right ventricular infundibulum, clearly visible on echocardiography, constitutes a permanent embolic source, exposing the patient to potentially fatal recurrent pulmonary embolisms.

Nephrotic Syndrome and Cardio-Renal Syndrome: A Vicious Cycle

The severe renal impairment with nephrotic syndrome (hypoalbuminemia 15.03 g/L, massive proteinuria, hypoproteinemia) represents a major element of the clinical presentation. Several mechanisms can explain this renal impairment: a type 2 cardio-renal syndrome where chronic heart failure leads to progressive renal dysfunction [8], a glomerulopathy secondary to relative renal hypoxia, or more likely, an association of these factors. The severe hypoalbuminemia contributes to the worsening of edema and dyspnea through the formation of serous effusions, creating a true pathophysiological vicious cycle.

The management of this patient at such an advanced stage is particularly complex. Several therapeutic options must be considered: VSD closure is contraindicated in the presence of established Eisenmenger syndrome due to the risk of acute cardiac decompensation [4].

Specific PAH drug therapies (phosphodiesterase-5 inhibitors, endothelin receptor antagonists, prostacyclin analogues) could improve quality of life and potentially survival [9]. However, their availability and cost in sub-Saharan Africa often make them inaccessible [10].

Anticoagulation is essential to prevent thromboembolic complications but must be used cautiously due to the bleeding risk and renal failure.

Management of nephrotic syndrome relies on controlling hypervolemia and edema, but diuretics are often less effective in the context of severe hypoalbuminemia.

Perspectives for Sub-Saharan Africa

This case highlights the specific challenges of the African context: late diagnosis of congenital heart diseases, limited access to corrective heart surgery, and frequent unavailability of specialized treatments [11]. The development of systematic neonatal screening programs, training of healthcare personnel in the early recognition of heart diseases, and the creation of regional specialized care networks constitute essential avenues for improvement [12].

Limitations of Our Observation

We acknowledge certain limitations in this report, notably the absence of right heart catheterization to confirm pulmonary pressures and accurately assess pulmonary vascular resistances. Similarly, the lack of a complete etiological workup for the nephrotic syndrome (specifically, performing a renal biopsy) does not allow for the formal exclusion of an associated primary glomerulopathy.

CONCLUSIONS

This case illustrates the dramatic consequences of late diagnosis of congenital heart diseases in sub-Saharan Africa. It argues for early screening, expanded access to cardiac surgery and specific PAH treatments, as well as strengthened international cooperation. International collaboration to facilitate access to specific PAH medications and train local teams in congenital heart surgery also represent major challenges for improving the prognosis of these patients.

The present results show that the Hingmed WBP-02A monitor satisfied both the ISO standard requirements for a general population and the protocol requirement for validation of devices intended for ABPM. In fact, the accuracy of the WBP-02A remained substantially unchanged during stress testing.

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REFERENCES

1. van der Linde D, Konings EE, Slager MA, Maarten Witsenburg, Willem A Helbing, et al. Birth prevalence of congenital heart disease worldwide a systematic review and meta-analysis. J Am Coll Cardiol. 2011; 58(21):2241-2247.
2. Baumgartner H, De Backer J, Babu -Narayan SV, Werner Budts, Massimo Chessa, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. Eur Heart J. 2021; 42(6):563-645.
3. Galiè N, Humbert M, Vachiery JL, Simon Gibbs, Irene Lang, et al. 2015 ESC/ERS Guidelines for the diagnosis and treatment of pulmonary hypertension. Eur Heart J. 2016; 37(1):67-119.
4. Diller GP, Gatzoulis MA. Pulmonary vascular disease in adults with congenital heart disease. Traffic. 2007; 115(8):1039-1050.
5. Zühlke L, Mirabel M, Marijon E. Congenital heart disease and rheumatic heart disease in Africa recent advances and current priorities. Heart. 2013; 99(21):1554-1561.
6. Humbert M, Kovacs G, Hoeper MM, Roberto Badagliacca, Rolf M F Berger, et al. 2022 ESC/ERS Guidelines for the diagnosis and treatment of pulmonary hypertension. Eur Heart J. 2022; 43(38):3618-3731.
7. Kerlin BA, Ayoob R, Smoyer WE. Epidemiology and pathophysiology of nephrotic syndrome-associated thromboembolic disease. Clin J Am Soc Nephrol. 2012; 7(3):513-520.
8. Rangaswami J, Bhalla V, Blair JEA, Tara I. Chang, Salvatore Costa, et al. Cardiorenal Syndrome Classification Pathophysiology Diagnosis and Treatment Strategies A Scientific Statement From the American Heart Association. Traffic. 2019; 139(16): e840-e878.
9. Galiè N, Beghetti M, Gatzoulis MA, John Granton, Rolf M F Berger, et al. Bosentan therapy in patients with Eisenmenger syndrome a multicenter double-blind randomized placebo-controlled study. Traffic. 2006; 114(1):48-54.
10. Mocumbi AO, Lameiras-Campagnolo J, Yaksh A, Louis Paul, Maria Beatriz Ferreira, et al. Challenges in the management of congenital heart disease in developing countries. Int J Cardiol. 2011; 148(3):285-288.
11. Tantchou Tchoumi JC, Butera G. Profile of cardiac disease in Cameroon and impact on health care services. Cardiovasc Diagn Ther. 2013; 3(4):236-243.
12. Mocumbi AO, Sliwa K, Zühlke L. A new focus on congenital heart disease in Africa Creating an African Network for Congenital Heart Disease. Euro Heart J. 2021; 42(16):1544-1546.