JOURNAL OF RARE CARDIOVASCULAR DISEASES

A rare combination of dextrocardia with an arachnoid cyst of the posterior fossa: A case report

Muchtar Nora Ismail Siregar[1,2,*], Vickry H Wahidji[1,2], Muhammad Isman Jusuf[2,3] and Zuhriana K Yusuf[4]

[1]Department of Cardiology and Vascular Medicine, Faculty Medicine, Gorontalo State University, Indonesia
[2]Aloei Saboe General Hospital Gorontalo, Indonesia
[3]Department of Neurology , Faculty Medicine, Gorontalo State University, Indonesia
[4]Department of Pharmacology, Faculty Medicine, Gorontalo State University, Indonesia

Correspondance: (e-mail: noraismailsiregar@ung.ac.id).

Received 10 November 2024 Revised 13 April 2025 Accepted 30 April 2025 Published 25 May 2025

ABSTRACT **Background:** Dextrocardia, a rare condition occurring in about 1 in 12,000 births, involves the abnormal positioning of the heart. While the exact cause remains unclear, it is believed to result from disruptions during early cardiac development. This case highlights the uncommon association between dextrocardia and a posterior fossa arachnoid cyst, a combination not frequently reported in the literature. **Case summary:** A 25-year-old female presented with symptoms of exertional dyspnea, orthopnea, paroxysmal nocturnal dyspnea, and fatigue, alongside bilateral peripheral edema. Her physical examination revealed elevated jugular venous pressure, tachycardia, and a systolic murmur, with ascites and lower extremity edema. Chest X-ray showed dextrocardia and cardiomegaly, and a CT scan of the brain identified an arachnoid cyst. Laboratory tests confirmed mild anemia, with normal renal function and serum protein levels. The patient was diagnosed with congestive heart failure and started on appropriate pharmacologic therapy, but she chose not to pursue further treatment for the arachnoid cyst, opting for outpatient follow-up instead. **Discussion:** The potential genetic association between dextrocardia and arachnoid cysts underscores the importance of comprehensive genetic evaluation in patients with congenital anomalies. While both conditions may be asymptomatic, their co-occurrence may be part of a broader developmental syndrome that warrants further exploration. Understanding the genetic basis of these conditions could provide valuable insights into their pathophysiology, aid in diagnosis, and guide management strategies in affected individuals.

KEYWORDS dextrocardia, arachnoid cyst, genetics, case report

1. INTRODUCTION

Congenital heart disease (CHD) refers to structural abnormalities of the heart or blood vessels present at birth and is the most common type of congenital malformation, contributing significantly to morbidity and mortality. Dextrocardia, a rare condition occurring in about 1 in 12,000 births, involves the abnormal positioning of the heart [1]. While the exact cause remains unclear, it is believed to result from disruptions during early cardiac development. Recent studies have pointed to genetic factors, such as copy number variations (CNVs) and specific mutations, which may be linked to isolated dextrocardia and could increase susceptibility to other congenital defects, including arachnoid cysts of the central nervous system [2]. This case highlights an unusual and underexplored association between dextrocardia and an arachnoid cyst in the posterior fossa, a co-occurrence that has not been frequently described in the medical literature.

2. CASE PRESENTATION

A 25-year-old female presented with a one-year history of exertional dyspnea, orthopnea, and paroxysmal nocturnal dyspnea. She also reported associated generalized fatigue and bilateral peripheral edema. Notably, the patient had no significant medical history, including hypertension, diabetes mellitus, chronic kidney disease, or smoking. Additionally, she experiences frequent headaches. On physical examination, her blood pressure was 100/60 mmHg, heart rate 125 beats per minute, respiratory rate 28 breaths per minute, and jugular venous pressure (JVP) was elevated. Cardiac auscultation revealed regular S1 and S2 heart sounds, with a systolic murmur audible at the 6th intercostal space along the right anterior axillary line. Pulmonary examination revealed vesicular breath sounds with bilateral rhonchi. Examination also revealed ascites and bilateral lower extremity edema.

Laboratory tests revealed mild anemia (hemoglobin 9.0 g/dL), preserved renal function (eGFR 105 mL/min/1.73 m^2), and normal levels of serum protein, albumin, and globulin. The electrocardiogram findings are depicted in Figure 1. A Chest X-ray showed dextrocardia, cardiomegaly, and signs of pulmonary congestion (Figure 2). Echocardiography showed mildly reduced left ventricular ejection fraction 44%, left atrial enlargement, and moderate mitral regurgitation (Figure 3). A contrast-enhanced brain CT scan identified an arachnoid cyst in the posterior fossa (Figure 4).

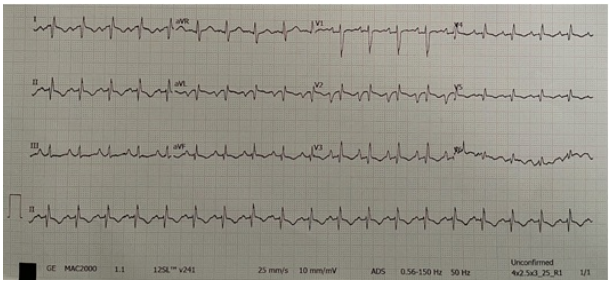

FIGURE 1. The electrocardiogram at presentation revealed global negativity in lead I, positive QRS with upright p wave and T wave in lead aVR, and absent R wave progression in precordial lead

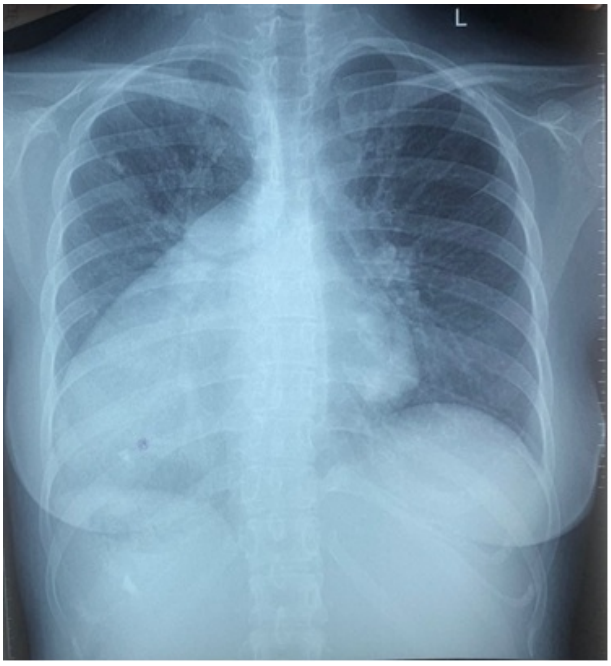

FIGURE 2. Chest X-ray showed dextrocardia with cardiomegaly, sign of pulmonary congestion

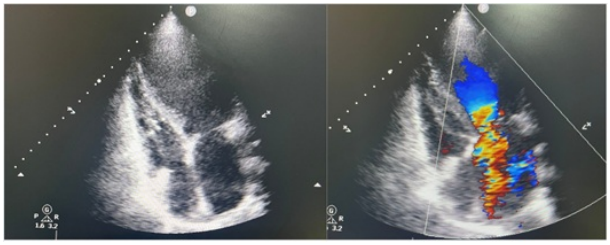

FIGURE 3. Echocardiography demonstrated moderate mitral regurgitation and left atrial enlargement

The patient was diagnosed with congestive heart failure with mildy reduced ejection fraction, dextrocardia, mitral regurgitation, arachnoid cyst and initiated intravenous diuretics with furosemide 120 milligrams per day, angiotensin-converting enzyme inhibitors, mineralocorticoid receptor antagonists, and beta-blockers. Although she was counseled regarding the management of the posterior fossa arachnoid cyst, the patient opted against further intervention. The patient underwent treatment for a duration of 5 days while maintaining stable hemodynamic conditions. She was subsequently discharged with instructions for routine follow-up in the outpatient clinic.

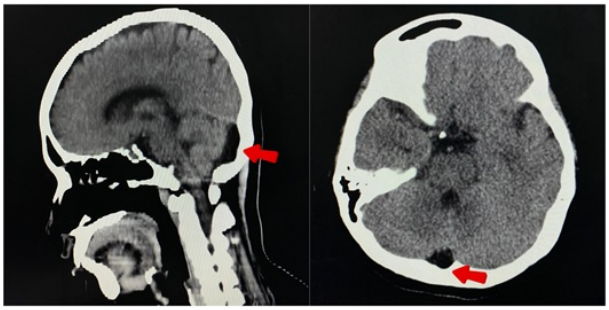

FIGURE 4. Contrast-enhanced brain CT scan identified an arachnoid cyst in the posterior fossa

3. DISCUSSION

This case is the first to document an unusual and underreported connection between dextrocardia and a posterior fossa arachnoid cyst, a co-occurrence infrequently reported in the literature. Although both dextrocardia and arachnoid cysts are rare on their own, their concurrent development in an otherwise young patient poses intriguing questions about possible embryologic and genetic relationships that might give rise to these malformations. Since both conditions are developmental processes that occur early in fetal life, the potential for common genetic or developmental mechanisms should be explored.

3.1 Dextrocardia: A Genetic and Developmental Perspective

Genetically, dextrocardia can be caused by disruptions in the left-right asymmetry signaling pathway during embryogenesis. The most important molecular pathways implicated in this process are sonic hedgehog (SHH), lefty-1, and nodal pathways, which are essential for the determination of normal leftward rotation and asymmetrical development of internal organs. Mutations in these gene or the pathway they control can result in situs anomalies, such as dextrocardia. In addition, dysfunction of cilia has been associated with dextrocardia in conditions like primary ciliary dyskinesia and Kartagener syndrome, both of which involve defective ciliary movement that compromises normal rotation of the organs during embryogenesis [2], [3]. Notably, investigation into the genetic etiology of situs anomalies continues, and recent evidence indicates that copy number variations (CNVs) and certain genetic mutations may play a role in isolated dextrocardia, potentially conferring increased susceptibility to other malformations, such as those of the central nervous system (CNS), such as arachnoid cysts.

3.2 Arachnoid Cysts in the Posterior Fossa: Genetic Underpinnings

Arachnoid cysts (ACs) are collections of cerebrospinal fluid (CSF) in the central nervous system lined with thin arach-

noid membranes contiguous with normal adjacent arachnoid containing hyperplastic arachnoid cells, excess collagen, and absence of normal spider-like trabeculations. The majority of AC patients are found to be asymptomatic, with 5%–12% of AC patients being symptomatic [4]. The precise etiology of arachnoid cysts is not entirely clear, but they are believed to be due to splitting or resorption failure of the arachnoid membrane during early brain development. Though the majority of arachnoid cysts are sporadic, certain IACs are believed to be secondary to gestational ischemic, traumatic, or infectious insult [5]. Experiments have shown correlations with a number of genetic syndromes involving arachnoid cysts, such as Ehlers-Danlos syndrome, Marfan syndrome, and neurofibromatosis type 1, all with underlying connective tissue defects and tissue development abnormalities. In addition, mutations in structural protein components that impair the integrity of the arachnoid layer of the brain or interruption of neurogenetic pathways may render individuals susceptible to cyst formation [6].

An enhanced prevalence of ACs has been reported in various syndromes hitherto. The occurrence of arachnoid cysts in patients with other congenital malformations is suggestive of a potential syndromic association, with partial evidence favoring common genetic pathways between CNS malformations and organ laterality defects. For instance, primary ciliary dyskinesia (PCD), a disorder well-documented to lead to dextrocardia, has been linked with cystic cerebral lesions, including arachnoid cysts, in a few instances [7]. This provokes the speculation that a common genetic defect in left-right patterning or cilia function might underlie the association between the two, both dextrocardia and arachnoid cysts.

3.3 Clinical Implications and Future Research

The possible genetic correlation between dextrocardia and arachnoid cysts makes it all the more critical that patients with congenital anomalies undergo thorough genetic assessment. Although both are likely to be asymptomatic, together they may represent part of an underlying developmental syndrome that should be investigated. The identification of overlapping genetic pathways not only might enhance our knowledge of these conditions but also might inform clinical management, especially in those who also have other associated congenital anomalies.

There will need to be further investigation into the genetic causes of dextrocardia and arachnoid cysts to determine possible connections as well as to discover the particular mutations or pathways that are responsible for their development. Further, the use of genomic sequencing in uncovering rare mutations would assist in developing a more accurate explanation of the genetic factors involved in the concomitant occurrence of these two diseases.

4. CONCLUSION

This case of a 25-year-old female with dextrocardia and an incidental posterior fossa arachnoid cyst presents an intriguing clinical scenario that may suggest a potential genetic or embryological connection. Although direct evidence linking dextrocardia with arachnoid cysts is limited, the possibility of shared genetic defects affecting left-right asymmetry and CNS development warrants further investigation. Understanding the genetic basis of these conditions could provide valuable insights into their pathophysiology, aid in diagnosis, and guide management strategies in affected individuals.

DATA AVAILABILITY

The data underlying this article will be shared on reasonable request to the corresponding author.

ABBREVIATIONS

eGFR: estimated Glomerular Filtration Rate
CT scan: Computerized Tomography
CNVs: Copy Number Variations
CNS: Central Nervous System
ACs: Arachnoid Cyst
CFS: Cerebrospinal Flui
PCD: Primary Ciliary Dyskinesia

COMPETING INTERESTS

The authors declare that they have no competing interests.

AUTHORS' CONTRIBUTIONS

Muchtar Nora Ismail Siregar, as the lead author, conducted the medical screening, designed the study, and drafted the manuscript. Vickry H. Wahidji contributed to patient review and investigation. Muhammad Isman Jusuf participated in patient review and investigation, as well as manuscript review and editing. Zuhriana K. Yusuf made substantial contributions to the conception and design of the study and was involved in reviewing and editing the manuscript.

ACKNOWLEDGEMENTS

The authors would like to thank all the staff of the Department of Cardiology and Vascular Medicine, Gorontalo State University, for their support

CONSENT

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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