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Case Report

Beyond cyanosis: paradoxical cerebral embolism in a pediatric patient with Ebstein's anomaly: A case report.

Fred Mwanikha[1], Ampuriire Nyakubaho[1,2], Harriet Nalubega Kisembo[1].
[1]Department of Radiology, Ernest Cook University (ECU).
[2]Department of Anatomy, Ernest Cook University (ECU).

ABSTRACT

Introduction:

Ebstein's anomaly is a rare congenital heart disease characterized by apical displacement of the tricuspid valve leaflets, accounting for approximately 0.5% of congenital cardiac defects. Associated intracardiac shunts may create a pathway for paradoxical embolism, an infrequently documented cause of arterial ischemia, particularly cerebral infarction.

Case Presentation:

This case report documents a 3-year, 7-month-old female with no significant prior history who presented with acute-onset seizures and right-sided spastic hemiparesis. Echocardiography confirmed Ebstein's anomaly with severe tricuspid regurgitation, atrialization of the right ventricle, and a ventricular septal defect. Brain CT demonstrated multiple infarcts within the left middle cerebral artery territory. In the absence of other embolic sources, paradoxical embolism through a right-to-left shunt was deemed the causative mechanism.

Conclusion:

Symptomatic paradoxical embolism is exceedingly rare in early childhood and may represent the initial manifestation of undiagnosed congenital heart disease. This case highlights the critical role of echocardiography in identifying the underlying cardiac anomaly and emphasizes the importance of considering paradoxical embolism in young patients presenting with cryptogenic stroke.

Take-away lessons:

Echocardiography is indispensable in pediatric cryptogenic stroke to exclude right-to-left shunts. Paradoxical embolism should be considered even in the absence of clinically evident venous thrombosis. Early recognition of structural heart disease in acute neurological presentations can prevent recurrent embolic events.

Keywords: *Paradoxical embolism; Ebstein's anomaly; Cerebral infarction; Echocardiography; Computed Tomography; Congenital heart disease.*
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Corresponding author: *Ampuriire Nyakubahoa*
Email: *nyakubaho7@gmail.com / ampuriire.nyakubaho@ecu.ac.ug*
Department of Anatomy and Radiology, Ernest Cook University (ECU), Mengo, Kampala, Uganda.
https://orcid.org/0009-0006-9035-302X

INTRODUCTION

Ebstein's anomaly is an uncommon congenital cardiac malformation, constituting about 0.5% of congenital heart diseases. It is defined by the displacement of the tricuspid valve leaflets from the atrioventricular ring into the right ventricle, leading to atrialization of the proximal ventricular portion and varying degrees of right ventricular dysfunction. (1,2). The clinical spectrum is broad, ranging from severe neonatal disease to asymptomatic survival into adulthood. (2,3).

A potential, serious complication is paradoxical embolism, which can occur when a right-to-left intracardiac shunt exists, such as an atrial or ventricular septal defect. (4,5). This allows venous thromboemboli to bypass the pulmonary circulation and enter the systemic arterial system, potentially causing cerebral, coronary, or peripheral infarction. Paradoxical embolism is a rarely confirmed diagnosis, often presumptively based on the triad of arterial embolism without a left-sided source, evidence of venous thrombosis, and the presence of an intracardiac shunt. (4,6). While echocardiography is the primary modality for diagnosing Ebstein's anomaly and associated defects (1,7), advanced imaging like magnetic resonance angiography (MRA) can offer superior assessment of cardiac anatomy and potential thrombus (8). This report presents a unique pediatric case where acute cerebral infarction from a paradoxical embolism was the inaugural symptom of previously undiagnosed Ebstein's anomaly.

CASE PRESENTATION

A 3-year, 7-month-old girl was admitted to a National Referral Hospital following a sudden generalized tonic-clonic seizure and loss of speech. Over the preceding

week, she had developed progressive right-sided body weakness, culminating in an inability to walk. Her developmental history was unremarkable until one year prior, when a gradual decline in physical activity was noted. There was no history of fever, trauma, cyanosis, or known chronic illness. Family history was non-contributory.

Examination revealed a semi-conscious child (Glasgow Coma Scale 11/15) with right central facial palsy and spastic hemiparesis. Cardiovascular assessment identified a grade 3/4 systolic murmur at the right 3rd-4th intercostal spaces. Routine laboratory studies were within normal limits, excluding an infectious or hematologic cause.

IMAGING FINDINGS

Echocardiography demonstrated classic features of Ebstein's anomaly: apical displacement of the septal and posterior tricuspid valve leaflets, atrialization of the proximal right ventricle, severe tricuspid regurgitation, and a ventricular septal defect (VSD) (1,7). Right ventricular volume was increased (5.6 cm^3). (Figure 1a and 1b).

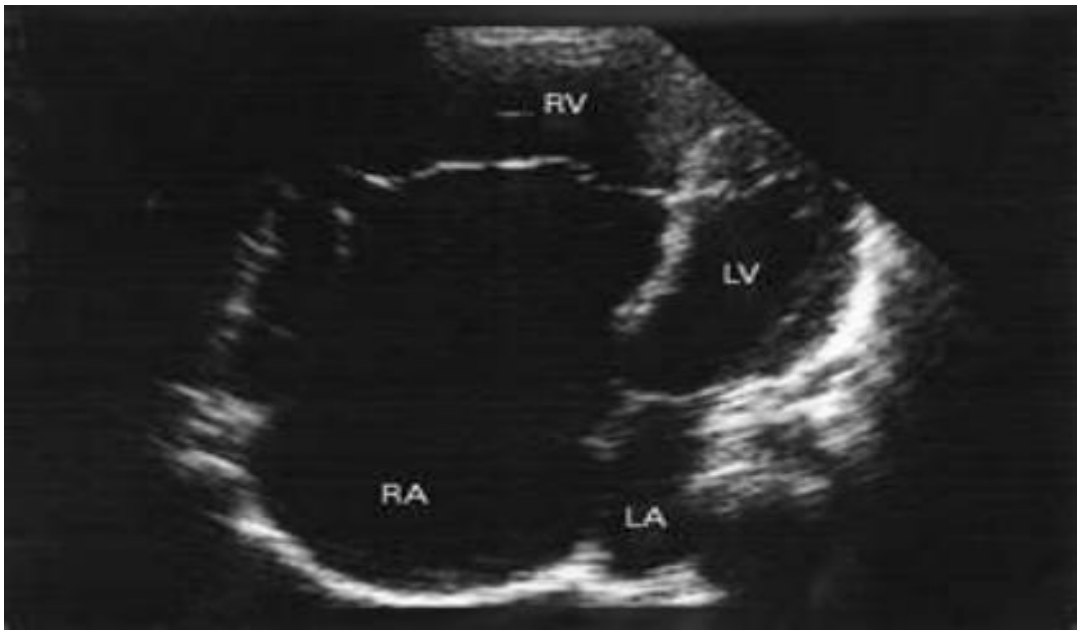

Figure 1a: *Apical four-chamber view showing apical displacement of the septal and posterior tricuspid valve leaflets, leading to atrialization of the right ventricle with a variable degree of malformation and displacement of the anterior leaflet.*

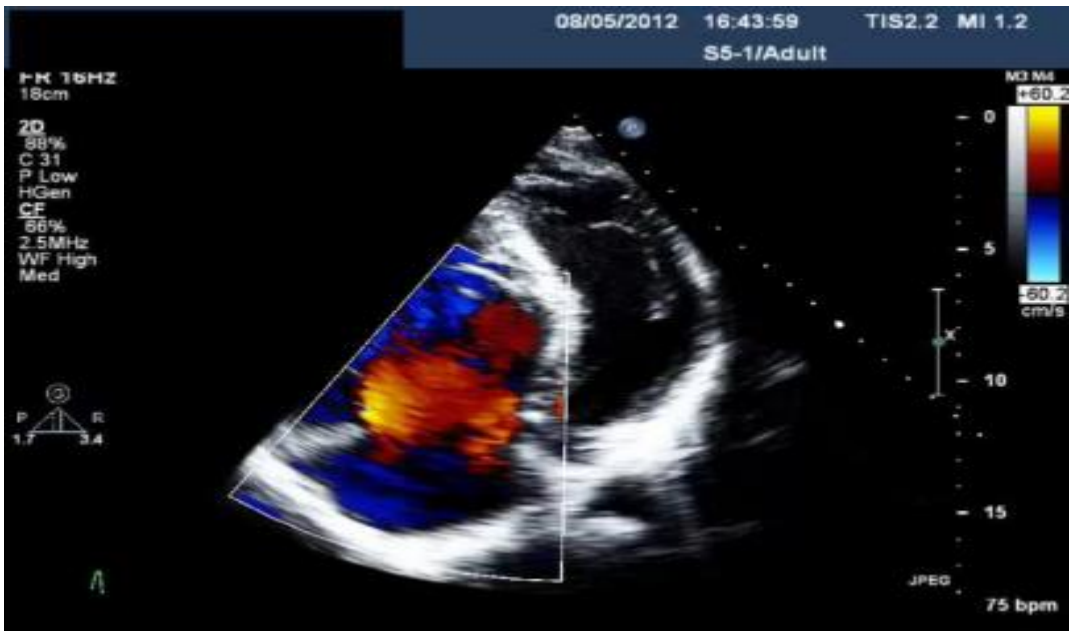

Figure 1b*: Colour doppler showing showing an associated ventricular defect causing high velocity in a highly situated right atrium.*

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Brain Computed Tomography (CT) showed a hypodense, contrast-enhancing lesion in the left parietal and occipital lobes, consistent with acute infarction in the M1/M2 territory of the left middle cerebral artery. (Figure 2a,2b and 2c)

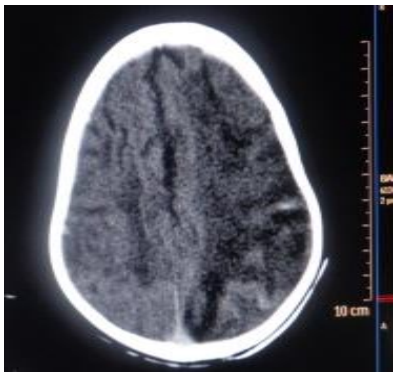

***Figure 2a**: Axial non contrasted brain CT scan showing an ill-defined hypodense lesion in the left parietal lobe.*

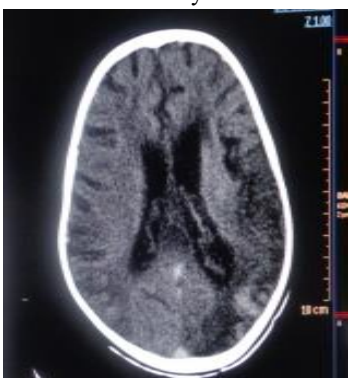

***Figure 2b**: Axial non contrasted helical brain CT scan showing the extension of the lesion into the occipital lobe*

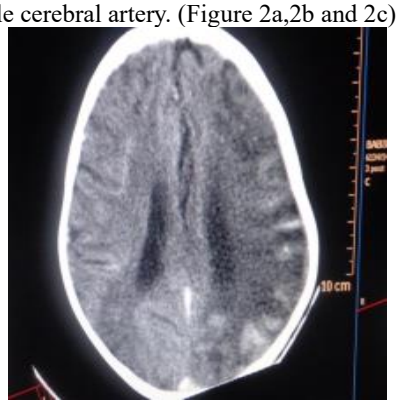

***Figure 2c**: Axial contrasted helical brain CT scan showing a luxurious enhancing lesion in the left parietal region.*

Given the imaging findings of a significant right-to-left shunt (via the VSD) and cerebral infarction without an alternative embolic source, a diagnosis of paradoxical embolism complicating Ebstein's anomaly was established. The patient was commenced on anti-failure therapy and referred for cardiology follow-up and surgical planning.

DISCUSSION

This case exemplifies a rare and life-threatening complication of Ebstein's anomaly, paradoxical embolism, causing cerebral infarction presenting in an unusually young child. The diagnosis of Ebstein's anomaly relies heavily on echocardiography, which effectively delineates the abnormal tricuspid valve morphology, atrialized right ventricle, and associated defects like VSD. (1,7,9). In this patient, echocardiography was instrumental in identifying the structural heart disease and the conduit for embolic paradox.

Paradoxical embolism, while a known entity, is an infrequently diagnosed cause of stroke, particularly in pediatrics (4,6). The diagnosis is often presumptive, based on clinical reasoning, as formal visualization of a thrombus traversing a shunt is uncommon. (4). The patient discussed here presented with the classic diagnostic dilemma: an arterial ischemic event without a left-heart source, in the setting of a proven intracardiac right-to-left shunt (VSD) (4,6). Although no deep vein thrombosis was clinically evident, the predisposition for stasis in the abnormally structured right heart in Ebstein's anomaly provides a plausible source for thrombus formation. (5,10).

The uniqueness of this case lies in the patient's age and presentation. While long-term survival into adulthood is possible with Ebstein's anomaly, significant morbidity and mortality often relate to heart failure or arrhythmias (2, 6). Symptomatic paradoxical embolism as the initial manifestation in a toddler is exceptionally uncommon and underscores the aggressive potential of this anomaly even in early life. This presentation aligns with the literature, noting that complications like paradoxical embolism can occur, but highlights their dramatic role as a sentinel event. (5,10).

Advanced cardiac imaging, particularly MRA, is recognized as superior to echocardiography for comprehensive anatomical evaluation and can be crucial for detecting intracardiac thrombi. (8). However, in resource-limited settings, echocardiography remains the vital first-line tool, as demonstrated here. Brain CT was sufficient to confirm the ischemic insult, guiding acute management.

TAKE AWAY LESSONS

1. Echocardiography is the first-line imaging modality for detecting right-to-left shunts in pediatric cryptogenic stroke and should be performed early in the diagnostic workup.
2. Paradoxical embolism should be suspected when arterial infarction occurs in the setting of an intracardiac shunt, even without demonstrable venous thrombosis.
3. Brain CT readily identifies territorial arterial infarctions and, when combined with echocardiographic findings, provides compelling circumstantial evidence for a paradoxical embolic mechanism.
4. Ebstein's anomaly with associated septal defects confers a tangible risk for systemic embolization, warranting prompt cardiology and

neurosurgical/neurology multidisciplinary evaluation.

CONCLUSION

This report details a distinctive case of Ebstein's anomaly in a young child, unveiled by catastrophic cerebral infarction due to paradoxical embolism. It reinforces several key concepts: Ebstein's anomaly has a variable and potentially severe natural history. (2,3); paradoxical embolism is a critical, albeit rare, complication that can be the initial presenting symptom (4–6); and echocardiography is the cornerstone for diagnosing the underlying cardiac defect (1,7). Clinicians must maintain a high index of suspicion for right-to-left shunts in young patients with cryptogenic stroke, as timely cardiac evaluation can reveal treatable structural heart disease and prevent further embolic events.

ABBREVIATIONS

1. CT - Computed Tomography
2. MRA - Magnetic Resonance Angiography
3. MRI - Magnetic Resonance Imaging
4. VSD - Ventricular Septal Defect
5. CBC - Complete Blood Count
6. GGT - Gamma-Glutamyl Transferase
7. ALT - Alanine Transaminase
8. ALP - Alkaline Phosphatase
9. WBC - White Blood Cell Count
10. HB - Hemoglobin
11. PLT - Platelet Count
12. GCS - Glasgow Coma Scale
13. HU - Hounsfield Units

DECLARATIONS.

Author Contributions

All the authors contributed to the drafting of the above manuscript. MF came up with the concept idea and case, AN contributed to the final drafting, grammar check, and literature review, and KN contributed to the discussion as well as supervising the whole drafting process.

Author biography

Fred Mwanikha is a Radiology Resident at Ernest Cook University.

Ampuriire Nyakubaho is an Assistant Lecturer at the Department of Anatomy and is also the Chief Radiology Resident at Ernest Cook University. He is also an early-career researcher with several publications and more currently under peer review in various journals.

Harriet Nalubega Kisembo is a Senior Consultant Radiologist and the head of Clinical and Professional Development at Ernest Cook University. She is a seasoned researcher with several publications and recently successfully defended her PhD in 2025.

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Conflict of interests.

The authors declare no conflict of interest.

Data Availability Statement

The data that supports the findings of this case report are available from the corresponding author upon reasonable request.

Participant consent

The participant was informed about the need to use the findings of the scan for learning, education, and publishing purposes, and consent was granted.

Ethics approval

Informed consent was obtained from the subject for this publication.

Source of finding

The authors receive no funding for the publication.

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