International Journal of Community Medicine and Public Health
Adiody S et al. Int J Community Med Public Health. 2026 Jul;13(7):3927-3930
https://www.ijcmph.com
pISSN 2394-6032 | eISSN 2394-6040

DOI: https://dx.doi.org/10.18203/2394-6040.ijcmph20262301

Case Report

A rare constellation of findings in VACTERL association

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Received: 21 April 2026
Accepted: 22 June 2026

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ABSTRACT

VACTERL (Vertebrae, anus, cardiac system, tracheo-esophageal fistula, renal system, and limbs) association is a heterogeneous constellation of congenital anomalies involving at least three of six characteristic components: vertebral, anorectal, cardiac, tracheo-esophageal, renal, and limb defects. We report a rare presentation in a 3-year-old girl who exhibited anorectal malformation, vertebral scoliosis, and complex cardiac abnormalities, including dextrocardia, bilateral superior vena cavae, and patent ductus arteriosus. She presented with chronic cough, recurrent respiratory infections, and wheezing. Clinical examination and imaging confirmed the anomalies, and echocardiography further delineated her cardiac defects. The constellation of findings met diagnostic criteria for VACTERL association. She improved with medical management and continues under multidisciplinary follow-up. This case highlights the wide phenotypic variability of VACTERL and underscores the importance of comprehensive systemic evaluation, especially when atypical cardiac or laterality abnormalities are present. Early recognition and coordinated long-term care are essential to optimize outcomes and identify additional anomalies that may emerge over time.

Keywords: VACTERL association, Dextrocardia, Bilateral superior vena cava, Anorectal malformation, Congenital anomalies

INTRODUCTION

The VACTERL association is a well-recognized pattern of non-random congenital anomalies involving vertebral defects, anorectal malformations, cardiac anomalies, tracheo-esophageal defects, renal anomalies, and limb abnormalities. A diagnosis is typically considered when at least three of these core features are present, reflecting its status as a developmental association rather than a defined syndrome.[1] The estimated birth prevalence ranges from 1 in 10,000 to 1 in 40,000 live births, although variability in diagnostic criteria contributes to disparities across studies.[1] Population-based European data have more recently suggested a prevalence of approximately 0.5-0.6 per 10,000 live births.[2] The etiology of VACTERL association remains poorly understood. Evidence suggests a multifactorial origin involving genetic susceptibility, perturbations during early embryonic development, and potential environmental or maternal influences.[3] Although familial clusters and chromosomal aberrations have been reported, most cases occur sporadically, with no identifiable inheritance pattern.[3] The heterogeneity in clinical presentation further complicates etiologic investigations, as patients often exhibit different combinations of anomalies, with many presenting with only three or four components of the association.[4] Cardiac anomalies are reported in approximately 40-80% of VACTERL cases and may range from simple septal defects to more complex structural malformations.[5] However, dextrocardia and bilateral superior vena cavae remain relatively rare within

this population. Their presence may complicate perioperative planning, anaesthesia, and long-term care.

The present case describes a 3-year-old girl with anorectal malformation, vertebral anomaly (scoliosis), and multiple cardiac abnormalities including dextrocardia, bilateral superior vena cavae, and PDA. Although she did not exhibit tracheo-esophageal, renal, or limb defects, the combination of anomalies fulfils diagnostic criteria for VACTERL association. This report highlights the phenotypic variability of the condition, emphasizes the importance of thorough systemic evaluation, and contributes to the limited literature describing unusual cardiac presentations within the VACTERL spectrum.

CASE REPORT

A 3-year-old female child presented to the pulmonology outpatient department with a 2-month history of persistent cough. There was no history of fever, but the parents reported intermittent wheezing episodes and recurrent respiratory tract infections since infancy.

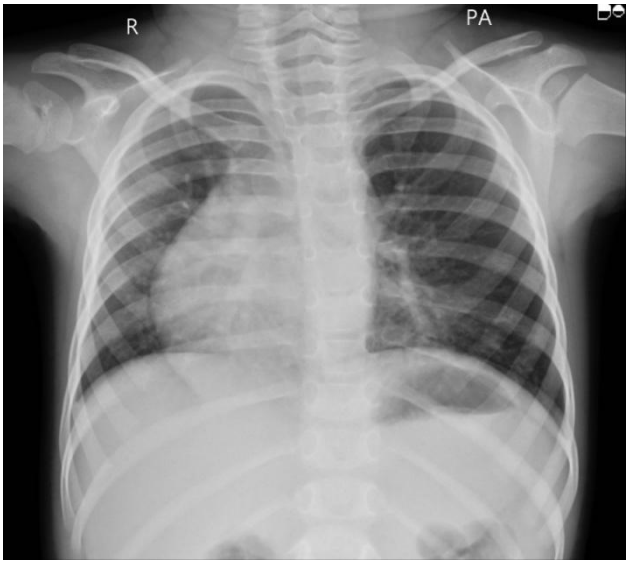

Figure 1: Chest radiograph demonstrating dextrocardia with the cardiac apex oriented toward the right hemithorax, along with associated vertebral scoliosis.

She was born at term via caesarean section and was diagnosed at birth with an anorectal malformation, for which she underwent an anterior sagittal anorectoplasty (ASARP) shortly after delivery. On examination, the child was hemodynamically stable, weighing 14 kg. Auscultation revealed bilateral wheeze. A chest radiograph showed dextrocardia and associated scoliosis (Figure 1). Routine blood investigations were within normal limits. She was treated with oral corticosteroids, antibiotics, antihistamines, a leukotriene antagonist, and inhaled corticosteroid therapy (ICS), with clinical improvement noted during follow-up. Cardiology evaluation was sought in view of dextrocardia on chest X-ray, to further characterize her cardiac findings. Transthoracic echocardiography (2D ECHO) demonstrated dextrocardia, bilateral superior vena cavae (SVC) (Figure 2), a small 2mm size patent ductus arteriosus (PDA) with a left-to-right shunt (gradient 70/16 mm Hg) (Figure 3), mild tricuspid regurgitation, and mild dilation of the left atrium and left ventricle.

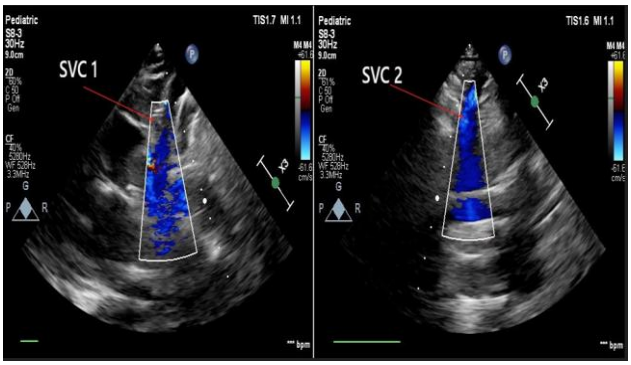

Figure 2: Transthoracic echocardiography (2D ECHO) showing bilateral superior vena cavae.

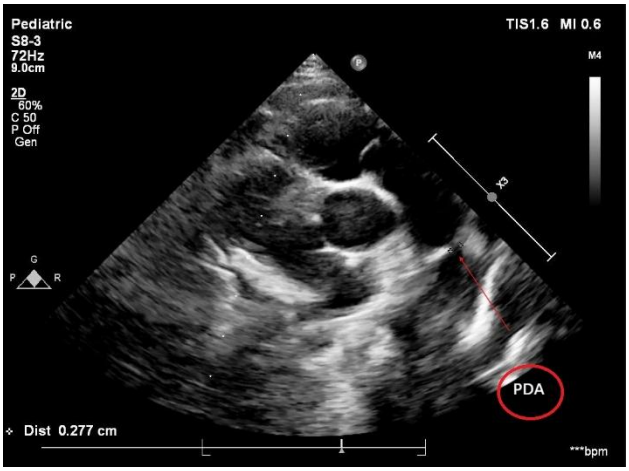

Figure 3: Transthoracic echocardiography (2D ECHO) showing a small (2 mm) patent ductus arteriosus (PDA).

Considering the constellation of congenital anomalies—including anorectal malformation, vertebral anomaly (scoliosis), and cardiac defects—the child was diagnosed with VACTERL association. She was advised computed tomography (CT) of chest to check for tracheoesophageal fistula, but parents were not willing to do the CT scan. She continues to remain stable and is under regular outpatient follow-up with multidisciplinary care coordination.

DISCUSSION

This case illustrates several important aspects of VACTERL association, beginning with its inherently heterogeneous presentation. Traditional diagnostic criteria

rely on the presence of at least three of the six characteristic anomalies: vertebral, anorectal, cardiac, tracheo-esophageal, renal, and limb defects.[1] Our patient demonstrated vertebral scoliosis, anorectal malformation, and complex cardiac anomalies, satisfying the minimal requirements. Many patients described in the literature similarly present with only partial involvement, typically three or four features, underscoring the broad phenotypic spectrum of VACTERL.[4]

Cardiac involvement represents one of the major components of the association, with congenital heart disease reported in 40-80% of patients.[5] The most frequently described defects include ventricular septal defects, atrial septal defects, and tetralogy of Fallot.[5] In contrast, dextrocardia and bilateral superior vena cavae—as seen in our case—are far less common. Published case series examining cardiac variability in VACTERL note that positional and laterality defects remain rare, making this presentation clinically noteworthy.[5] Such variants may have implications for interventions, as unusual venous return patterns can complicate central line placement, cardiac catheterization, and surgical approaches.

The presence of additional non-classical abnormalities is well documented in VACTERL patients. Many exhibit rib anomalies, genitourinary malformations, or airway abnormalities not included in the core diagnostic criteria.[4] These extracardiac, extra-systemic anomalies are believed to reflect disturbances in blastogenesis—early embryonic development during which multiple organ systems arise concurrently. Dextrocardia, although not part of the classical definition, may represent such a perturbation of embryonic left–right axis formation.

Regarding pathogenesis, the etiology of VACTERL remains incompletely defined. A multifactorial model is widely accepted, with proposed contributors including genetic disruption of mesodermal development, environmental exposures, maternal diabetes, and assisted reproductive technologies.[2,3] Reports of chromosomal anomalies and rare single-gene mutations associated with VACTERL-like phenotypes provide evidence for a genetic component, yet no single unifying genetic mechanism has been identified.[3] The majority of cases—including ours occur without identifiable risk factors.

From a clinical standpoint, infants diagnosed with anorectal malformations, such as our patient, have been shown to have a significantly higher prevalence of congenital heart disease when VACTERL association is present.[6] These children often require earlier cardiac evaluation and, in some cases, surgical correction. Although our patient's cardiac lesions were hemodynamically mild, their structural complexity—particularly dextrocardia and bilateral SVC necessitates continued cardiology surveillance. Even small PDAs may warrant follow-up to assess for spontaneous closure or progression. The vertebral anomaly (scoliosis) is another important consideration. Vertebral defects occur in up to 60-80% of VACTERL cases and may have long-term consequences on posture, respiratory function, and growth. Given our patient's recurrent wheezing and history of frequent respiratory infections, her scoliosis may further influence pulmonary mechanics, making collaboration between pulmonology and orthopaedics essential.

Because VACTERL cases frequently harbour additional occult anomalies, periodic reassessment is recommended. Renal anomalies, for example, may initially be subtle or asymptomatic, yet can have significant long-term implications if unrecognized. Likewise, subtle limb anomalies or functional impairments may only become apparent with age. Several published case reports stress the value of longitudinal follow-up for early identification of late-presenting anomalies.[7]

The unusual configuration of cardiac structures in our patient is particularly relevant for anaesthetic and procedural planning. Bilateral SVC can affect venous drainage patterns, complicating central venous access or cardiopulmonary bypass strategies. Thus, comprehensive anatomical evaluation is crucial for any future procedures, even if current cardiac function appears stable.

This case also underscores the importance of recognizing atypical presentations within the VACTERL spectrum. Reporting such variations enriches existing literature, expands the documented phenotypic spectrum, and helps clinicians anticipate potential abnormalities in patients with partial features. Moreover, documenting rare cardiac variants especially laterality defects may contribute to future efforts to better understand developmental mechanisms underlying VACTERL.

CONCLUSION

This case highlights an uncommon presentation of VACTERL association with anorectal malformation, vertebral scoliosis, dextrocardia, bilateral superior vena cavae, and PDA. It underscores the considerable phenotypic variability of the condition and emphasizes the need for comprehensive systemic evaluation even when only partial criteria are met. Long-term multidisciplinary monitoring is essential for detecting additional anomalies, guiding management, and anticipating procedural challenges related to complex cardiac anatomy. Reporting such atypical combinations contributes to a broader understanding of the developmental disturbances underlying VACTERL and enhances clinical recognition of its diverse manifestations.

ACKNOWLEDGEMENTS

The authors would like to thank the patient and her family for their cooperation and for providing consent to share

this case for academic purposes. The authors also acknowledge the contributions of the departments involved in the diagnosis and management of the patient, whose multidisciplinary support was essential in the care of this case.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Adiody S, Narayanan V, Kunnambath G, Viswambaran B. A rare constellation of findings in VACTERL association. Int J Community Med Public Health 2026;13:3927-30.