Bristol Publishers

Case Reports and Case Series in Cardiology Journal

Case Report | Open Access | Volume: 01 Issue: 02 (July-September 2025) Pages: 37-40

ISSN: 3107-6416 (Online) | DOI: doi.org/10.64874/crcscj.v1i2.2025.008

Fallot-type Double Outlet Right Ventricle and Associated Neurodevelopmental Delays in an 11-Year-Old Female in a Resource-Poor Setting: A Case Report

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Received: 12 May 2025; **Revised:** 20 July 2025; **Accepted:** 22 August 2025; **Published:** 25 September 2025

Academic Editor: Dr. Shreyas Samaga

Abstract

Background: Double Outlet Right Ventricle (DORV) is a cyanotic congenital heart disease in which both the pulmonary artery and the aorta arise from right ventricle; hence, via an often-present ventricular septal defect (VSD), oxygenated blood in left ventricle returns to right. When it has features that mimic Tetralogy of Fallot (TOF), there will also be overriding aorta, pulmonary stenosis, as well as right ventricular hypertrophy. DORV can be associated with neurodevelopmental delays, particularly in children from resource-limited settings.

Case Presentation: We present the case of 11-year-old female, the sixth of seven children born to subsistence farmers of Igbo tribe, who presented with lifelong symptoms of breathlessness on mild exertion, easy fatigability, poor academic performance, poor growth and lack of self-care. Physical examination revealed central cyanosis, reddish conjunctivae, grade 4 digital clubbing, microcephaly (OFC 48cm), and neurodevelopmental delays, including an unsteady gait, hypertonia in the lower limbs, and disorientation. Cardiovascular examination showed a pulse rate of 135 beats per minute and a pansystolic murmur (A pansystolic murmur can occur too due to VSD depending on the pressure gradient) at left lower sternal border. Echocardiography confirmed the presence of TOF-type DORV. Urgent blood test revealed polycythaemia. The patient's developmental delays were consistent with chronic hypoxemia arising from the congenital heart disease.

Conclusion: This case underscores importance of early diagnosis as well as intervention in children with TOF-type DORV, mainly in resource-limited settings. The neurodevelopmental delays seen in this patient draw attention to the need for multidisciplinary management approach that addresses both cardiac as well as neurological aspects of congenital heart disease in a resource-limited setting.

Keywords: Neurodevelopmental delay; Tetralogy of fallot; Double outlet right ventricle

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Introduction

DORV is a congenital heart defect characterized by abnormal origin of both pulmonary artery as well as aorta from right ventricle. It encompasses multiple subtypes that can clinically resemble other congenital heart conditions, including VSD, single ventricle physiology, transposition of great arteries, or TOF [1]. DORV is second most typical congenital heart defect in Nigeria, following TOF [2]. The Fallot-type DORV closely mimics TOF, sharing the hallmark features of VSD, overriding aorta, pulmonary stenosis, as well as right ventricular hypertrophy. These structural anomalies impair oxygenation, leading to cyanosis and clinical manifestations such as exertional dyspnea, poor feeding, and failure to thrive [3].

While DORV is primarily a cardiovascular disorder, it is also associated with extracardiac manifestations, these are the effects of cyanosis including neurodevelopmental delays and cognitive impairments. The relationship between chronic hypoxemia and neurodevelopment is complex, with children affected by cyanotic heart defects being at an elevated risk for neurodevelopmental disorders [4]. Although the association between congenital heart disease and developmental delays is well-documented, the severity and impact of neurological deficits can vary widely [5].

In this case report, we describe an 11-year-old female with TOF-type DORV who exhibited significant developmental delays, including microcephaly, poor academic performance, and challenges with self-care. This case underscores the importance of considering neurodevelopmental results in children with congenital heart disease, particularly in resource-constrained settings where access to early intervention services may be limited. Our report adds to the growing body of literature on the long-term outcomes of children with TOF-type DORV and emphasizes the need for comprehensive care that addresses both cardiac and developmental aspects of these patients' health.

Technical issues: A case report with images was intended, but retrieval of images was stalled by technical issues that affected the storage system in the echocardiography machine. When all efforts at retrieval failed, the need to go ahead with this very important and informative case report prevailed. However, the relevant details of the findings are presented here.

Case Presentation

11-year-old A.B., female, Igbo by tribe, born to subsistent farmers aged 56 (father) and 42 (mother) years old. Patient is the 6th of the parents' 7 children and presented with a lifelong history of breathlessness on mild exertion, easy fatigability, poor academic performance, poor growth and lack of self-care. Pregnancy and birth history were found to be unremarkable.

On examination, we found an acutely ill child with bilateral red conjunctiva, central cyanosis, and grade 4 digital clubbing. She had microcephaly with an OFC of 48 cm. There was an abnormal staggering gait and hypertonia in lower limbs. She was oriented in person and time, but not in place, and was unable to draw a person. Pulse rate was 135 beats per minute, with pansystolic murmur heard at lower left sternal border.

Investigations done showed polycythaemia (packed cell volume of 57%), while echocardiogram revealed laevocardia, right atrial and ventricular dilatation, right ventricular hypertrophy, large peri membranous VSD with right to left shunt. There was also a valvular pulmonary stenosis with moderate gradient. There were small-sized pulmonary arteries, normal coronary arteries, good systolic function. Patient had aortic overriding (75%) equivalent to double outlet right ventricle. Surgical correction was advised, while multivitamin syrup and propranolol were prescribed for conservative care. Saline pheresis was also done to reduce the polycythaemia and improve clinical outcome, and child was referred to child neurologist given the neurodevelopmental delay.

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Discussion

TOF-type DORV is a complex congenital heart defect with a spectrum of clinical manifestations primarily related to its cyanotic nature. In the case presented, the patient's clinical features, including central cyanosis, digital clubbing, and exertional dyspnea, are classic signs of TOF –type DORV. However, the accompanying neurodevelopmental delays, as evidenced by microcephaly, poor academic performance, and difficulties in self-care, provide an important dimension to the management and prognosis of this patient [6]. Besides, cyanotic congenital heart defects are known to cause more neurodevelopmental defects than the acyaonotic heart defects, hence, earlier screening with concomitant specialist neurological consultations is very vital [7].

Clinical Correlation:

The patient's presentation with cyanosis and polycythaemia aligns with the pathophysiology of TOF-type DORV, where decreased pulmonary blood flow leads to chronic hypoxemia [8]. The body compensates for this reduced oxygen saturation through increased red blood cell production, resulting in secondary polycythaemia. This chronic state of hypoxemia has been implicated in development of neurological sequelae, encompassing cognitive impairment as well as developmental delays, found in about 42 percent of patients with congenital heart diseases [9].

The neurological findings in this patient, such as microcephaly, hypertonia, and disorientation, may be multifactorial. Chronic hypoxemia is known to impair brain development and function, potentially leading to delayed milestones, learning difficulties, and behavioural issues. The degree of neurological impairment can vary depending on severity of hypoxemia, timing of surgical intervention, and the presence of other comorbid conditions.

Literature Review:

The association between congenital heart defects and neurodevelopmental delays is well-documented. Studies have shown that children with congenital heart defects are at increased risk for cognitive deficits, motor delays, and behavioural problems [8]. These impairments are thought to result from both the hemodynamic effects of the heart defect and the adverse effects of chronic hypoxemia on the developing brain [8]. In resource-limited settings, where access to timely surgical intervention and developmental support services may be constrained, these issues can be exacerbated, leading to poorer long-term outcomes.

Our patient's case is illustrative of the challenges faced in managing these patients in such environments. Despite the classical cardiac findings that led to the diagnosis, the neurodevelopmental aspects were more insidious, likely contributing to her poor academic performance and difficulties with self-care. This case underscores the need for a multidisciplinary approach to the management of children with this condition, encompassing not only cardiac care but also neurodevelopmental assessment and support [7].

Management and Prognosis:

The management of TOF traditionally focuses on surgical correction. The same applies to TOF–type DORV, which will require surgical intervention to correct the anomaly early. However, as demonstrated by this case, the timing of surgery and the preoperative state of the patient, particularly regarding neurological development, play crucial roles in the overall prognosis [10]. In the case of our patient, early intervention, including corrective surgery and comprehensive developmental support, would have been ideal. Unfortunately, late diagnosis and the possible lack of early neurodevelopmental interventions may have contributed to her current developmental status.

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Educational Value:

This case emphasizes the importance of early diagnosis and intervention in children with TOF - type DORV. It also highlights the critical need for ongoing neurodevelopmental monitoring and support, especially in resource-limited settings. Given the increasing survival rates of children with congenital heart defects due to advancements in surgical techniques, attention must now turn to improving the quality of life for these patients by addressing the long-term neurodevelopmental consequences of their condition.

Conclusions

In conclusion, the presented case of an 11-year-old female with TOF-type DORV and significant neurodevelopmental delays serves as a reminder of the multifaceted challenges faced by these patients. A holistic approach that integrates both cardiac and neurological care is essential for optimizing outcomes and ensuring that these children achieve their full potential.

Conflicts of Interest: The authors declare no conflict of interest.

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