Section: General Medicine

Case Report

AN ASYMPTOMATIC RARE CASE OF CYANOTIC CONGENITAL HEART DISEASE IN A 19-YEAR-OLD MALE: A CASE REPORT

Ankur Gupta[1], Jagjit Singh Bahia[2], Anshu Arora Bahal[3]

[13rd] Year PG Resident, Department of General Medicine, N C Medical College and Hospital, Panipat, Haryana, India.
[2]Professor, Department of General Medicine, N C Medical College and Hospital, Panipat, Haryana, India.
[3]Assistant Professor, Department of General Medicine, N C Medical College and Hospital, Panipat, Haryana. India.

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Corresponding Author:
Dr. Ankur Gupta,
3rd Year PG Resident, Department of General medicine, N C Medical College and Hospital, Panipat, Haryana, India.
Email: ankurgupta8401@gmail.com

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ABSTRACT

Background: Cyanotic congenital heart diseases (CCHDs) are complex structural defects typically diagnosed in early life due to overt clinical symptoms. However, late presentations, especially asymptomatic cases, are exceedingly rare. **Objectives:** To report an unusual case of an asymptomatic adult diagnosed incidentally with congenitally corrected transposition of the great arteries (CCTGA) and associated cardiac anomalies.
Materials and Methods: A 19-year-old male was referred for evaluation after being declared unfit during routine blood donation. A detailed history, physical examination, chest radiograph, ECG, and transthoracic echocardiography were performed. Due to poor imaging windows, the patient was referred to a tertiary care center for advanced evaluation.
Results: Echocardiography revealed dextrocardia, CCTGA, a large perimembranous ventricular septal defect (VSD), severe pulmonary stenosis, and severe tricuspid regurgitation, with preserved biventricular function. Despite these findings, the patient remained completely asymptomatic. He was referred for planned surgical correction.
Conclusion: This case demonstrates that CCTGA with severe associated anomalies can remain clinically silent well into adulthood. Incidental diagnosis in an asymptomatic patient underscores the importance of routine screening and clinical vigilance. Early detection is crucial to prevent complications and guide timely intervention.
Keywords: Cyanotic congenital heart disease; CCTGA; Dextrocardia; Asymptomatic presentation; Ventricular septal defect; Tricuspid regurgitation.

INTRODUCTION

Congenital heart disease (CHD) refers to structural abnormalities of the heart or great vessels that arise during fetal development and are present at birth. These defects can vary in severity and may affect the walls of the heart, the valves, or the arteries and veins near the heart. Broadly, CHDs are classified into two main categories: acyanotic and cyanotic heart diseases. Acyanotic defects typically involve left-to-right shunts, such as atrial or ventricular septal defects, while cyanotic defects involve right-to-left shunting, leading to the mixing of oxygen-poor blood with oxygen-rich blood and resulting in systemic hypoxemia and clinical cyanosis.[1,2]India bears a significant burden of congenital heart diseases, with an estimated incidence of 9 out of every 1,000 live births.[3] This translates to approximately 240,000 children born with CHD each year. Among these, cyanotic CHDs represent a smaller but more severe subset, demanding early recognition and intervention due to the risk of complications like heart failure, growth retardation, and death in undiagnosed cases.[3]One of the rarest forms of cyanotic CHD is Congenitally Corrected Transposition of the Great Arteries (CCTGA), accounting for less than 1% of all CHD cases.[4] CCTGA involves both atrioventricular and ventriculoarterial discordance, resulting in a physiologically corrected but anatomically abnormal circulation. While this "correction" can allow patients to remain asymptomatic for years, the

condition is often associated with additional cardiac anomalies like ventricular septal defects (VSD), pulmonary stenosis (PS), and tricuspid regurgitation (TR)—which may eventually manifest with heart failure, arrhythmias, or cyanosis.[5,6]

In this case report, we present a 19-year-old asymptomatic male incidentally diagnosed with CCTGA and associated cardiac defects, highlighting the importance of routine screening in detecting silent congenital heart diseases.

Case Presentation

A 19-year-old unmarried male, a first-year B.Tech student, was referred to the Department of General Medicine from the pathology unit at a tertiary care center after being declared unfit during routine blood donation screening. He was completely asymptomatic at the time of presentation, with no history of breathlessness, palpitations, chest pain, syncope, or fatigue. There were no cyanotic spells or recurrent respiratory infections during childhood, and the patient denied any limitation in physical activity.

Personal History

There was no past medical history of chronic illnesses such as diabetes mellitus, hypertension, bronchial asthma, tuberculosis, epilepsy, or coronary artery disease. The patient had never undergone surgery, taken long-term medications, or been exposed to radiation. Personal history revealed that he was a non-smoker, non-alcoholic, followed a vegetarian diet, and maintained normal appetite, sleep, and bowel-bladder habits. No addictive behavior was reported.The patient was the third of four siblings. All siblings and other family members were asymptomatic with no known history of congenital or acquired heart disease. There was no family history suggestive of genetic syndromes or cardiomyopathies.

General Examination

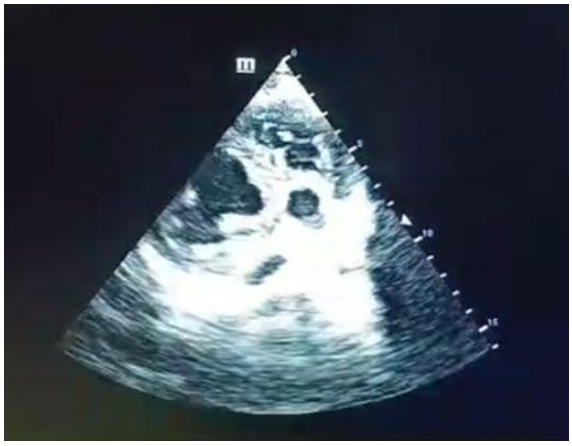
On general physical examination, the patient was alert, oriented, and cooperative. He appeared underweight with a height of 5 feet 11 inches and a weight of 52 kg, resulting in a BMI of 16.05 kg/m². Vitals were stable: blood pressure was 110/70 mmHg in the right arm and 114/76 mmHg in the left; pulse rate was 86 bpm, regular with good volume and character; and SpO_2 was 98% on room air. Clinical signs included central cyanosis and grade II digital clubbing. There was no pallor, icterus, lymphadenopathy, pedal edema, or raised jugular venous pressure.

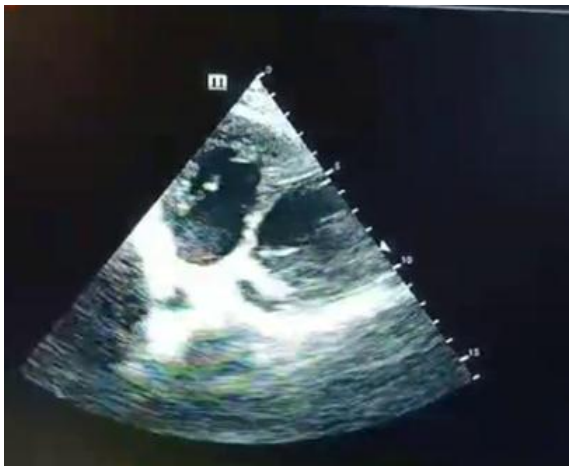
Systemic Examination

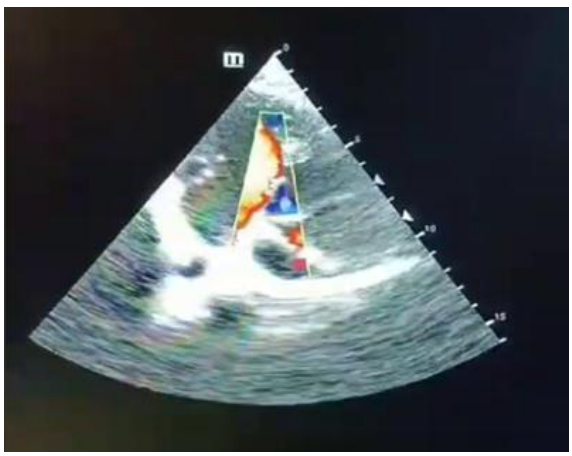
Cardiovascular examination revealed that the apical impulse was visible and palpable on the right side of the chest, suggestive of dextrocardia. No scars or engorged veins were noted. On auscultation, the heart rate was regular with normal first and second heart sounds, and a pansystolic murmur was appreciated best over the right parasternal area. No palpable thrill or abnormal heart sounds were noted. Other systemic examinations were within normal limits.

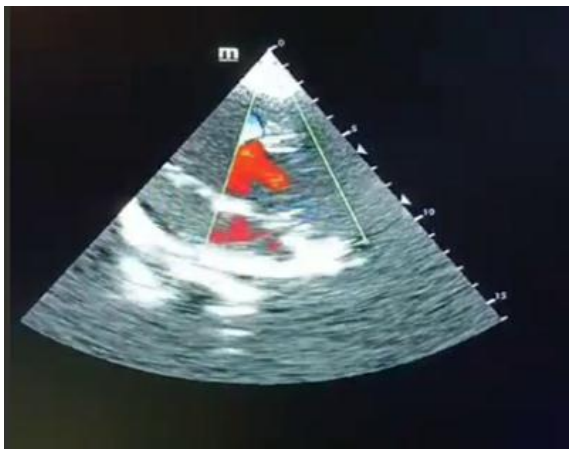
Investigations

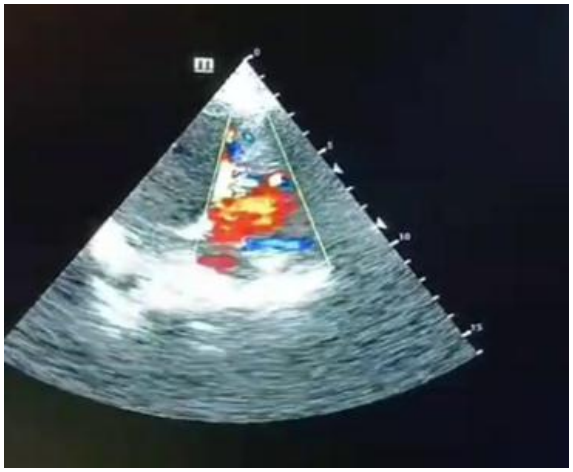
Laboratory investigations revealed a hemoglobin level of 10.7 g/dL, total leukocyte count of 8,600/mm³, and platelet count of 3.57 lakh/mm³. Liver and renal function tests were within normal limits. Electrocardiogram showed T wave inversion in leads I, aVL, and V1. Chest radiograph (PA view) demonstrated dextrocardia. Initial 2D transthoracic echocardiography at the tertiary care center revealed dextrocardia with a poor acoustic window, presence of a ventricular septal defect (VSD), normal ejection fraction (>55%), no valvular lesions, and no shunt reversal or regional wall motion abnormalities.











Further Evaluation and Management

In view of the structural anomalies and dextrocardia, the patient was referred to PGIMER, Chandigarh for detailed cardiac evaluation. Repeat echocardiography confirmed the diagnosis of Congenitally Corrected Transposition of the Great Arteries (CCTGA) along with a large perimembranous VSD, severe pulmonary stenosis, and severe tricuspid regurgitation. Biventricular function was preserved. Although the patient remained entirely asymptomatic, the presence of significant structural cardiac defects warranted corrective surgical planning, and he was referred for cardiothoracic intervention.

DISCUSSION

A rare presentation of a 19-year-old male diagnosed incidentally with CCTGA, large perimembranous VSD, severe pulmonary stenosis, and tricuspid regurgitation—despite being entirely asymptomatic. Unlike the findings of Varma et al,[7] and Rao et al,[8]where most CCHD cases present early with breathlessness, cyanosis, or recurrent infections, this patient had no such symptoms, suggesting a well-compensated physiology and slow disease progression.The structural findings align with existing literature. Varma et al,[7] reported VSD as the most common associated defect, while Rao et al,[8] and Zhao et al,[9] highlighted progressive tricuspid regurgitation in CCTGA due to systemic ventricular overload. However, the incidental discovery during blood donation screening is highly unusual and not reported in the compared studies, emphasizing the value of routine health assessments even in asymptomatic individuals.Imaging challenges due to dextrocardia, later clarified through tertiary-level echocardiography, mirror diagnostic difficulties noted by Zhao et al.[9] While the anatomical findings are consistent with known complications of CCTGA, the asymptomatic clinical profile and incidental detection make this case exceptional. It underscores the importance of considering congenital heart disease even in the absence of symptoms and supports the role of early diagnosis to guide timely intervention.

CONCLUSION

We concluded that CCTGA with significant associated defects can remain asymptomatic into adulthood, as demonstrated in this rare case. Early identification through incidental screening reinforces the importance of considering congenital anomalies even in asymptomatic individuals. This case broadens the clinical spectrum of CCTGA and supports the need for thorough cardiovascular evaluation in routine assessments.

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