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TETRALOGY OF FALLOT IN AN INDIAN CHILD

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ABSTRACT

Tetralogy of Fallot is a common congenital heart condition characterized by four primary features and several minor associated findings. The four primary elements consist of aortic override, a membranous ventricular septal defect, right ventricular hypertrophy, and obstruction of the right ventricular outflow tract. The patient's prognosis is determined by the severity of the obstruction in the right ventricular outflow tract. Literature documents cases of survival past the fifth decade, whether or not correction was applied. Cyanosis emerges during the first year of life if it is not evident at birth. When mild right ventricular outflow tract obstruction and a balanced ventricular septal defect prevent cyanotic spells, the condition is known as “Pink Tetralogy of Fallot.” We report a 12-year-old Indian girl with acyanotic pink tetralogy of Fallot, characterized by typical echocardiographic findings with mild aortic override.

KEYWORDS: *Tetralogy of Fallot (TOF), Overriding of Aorta, Right ventricular outflow obstruction, Syncopal attacks, Tet spells.*

INTRODUCTION

Tetralogy of Fallot, a prevalent cyanotic congenital heart defect, is defined by four primary characteristics: a ventricular septal defect, dynamic right ventricular outflow tract obstruction, an overriding aorta and right ventricular hypertrophy. The clinical manifestation and severity of this condition are determined by the extent of RVOTO, the relative pressures in the right and left ventricles, and the degree of aortic override of the VSD.

TOF -classical anatomy

Tetralogy of Fallot, occurring in approximately one out of every 3,500 live births, is a prevalent type of cyanotic congenital heart defect that represents 7–10% of all such cardiac anomalies.[1] Although Tetralogy of Fallot was identified as a cluster of defects in 1671, it received its name in 1888 following a case series that detailed the four primary anatomical and pathological characteristics: ventricular septal defect, right ventricular outflow tract obstruction, overriding aorta, and right ventricular hypertrophy.[2, 3]

Ventricular septal defect

This condition typically involves a considerable sized defect that does not impede blood flow across the ventricular septal defect. This defect typically occurs in the muscular and perimembranous areas of the ventricular septum, enabling blood to flow between the ventricles, specifically from right to left in classic cyanotic tetralogy of Fallot.

Outflow tract obstruction of the RV

A hallmark of TOF[3] is the blockage of pulmonary blood flow at the right ventricular outflow tract. When a ventricular septal defect is unrestrictive, escalating right ventricular outflow tract obstruction elevates right ventricular pressure, promotes right-to-left shunting, diminishes pulmonary blood flow, and results in hypoxaemia. When the patent ductus arteriosus is closed and collateral circulation is absent, the degree of hypoxemia at presentation correlates with the severity of right ventricular outflow tract obstruction, a characteristic feature of subvalvular obstruction seen in half of patients. This condition is typically dynamic and frequently stems from enlarged muscle bundles in the infundibulum. Alternatively, the blockage might occur at the pulmonary valve (10%) or further upstream (30%).[4] Pulmonary atresia refers to the total blockage of the right ventricular outflow tract.

Aortic Overriding

In TOF, the aorta primarily originates from the left ventricle, with only a minor contribution from the right ventricle. This state functions as the override.[3, 5] When the aorta arises mainly from the right ventricle, the condition is termed double outlet right ventricle (DORV), with its physiology dictated by the location of the ventricular septal defect and any accompanying outflow obstructions in the right or left ventricles.[3]

Hypertrophy of the RV

RV hypertrophy results from RVOTO, as elevated right ventricular pressure is required to sustain pulmonary blood flow, thereby modifying cavity dimensions and muscle mass, factors that remain significant following TOF repair.

Variants of TOF

TOF subtypes are primarily categorized based on the severity of cyanosis.[3] These are the most significant variants.

'Pink Fallot'

Children with this variant are acyanotic and have normal or near-normal oxygen saturations, with no or normal RVOTO.

From a physiological standpoint, the lesion functions as a considerable sized, non- restricted ventricular septal defect characterized by a left-to-right shunt. Patients may exhibit heart failure or other signs of a left-to-right shunt. While patients may initially exhibit heart failure or other signs of a left-to-right shunt, these symptoms often evolve as the child matures, increasingly resembling those of classic Tetralogy of Fallot.

Pulmonary atresia (15% of TOF)

As the most severe form, this variant features total pulmonary valve atresia, which completely blocks blood flow from the right ventricle into the pulmonary artery. Intracardiac mixing is crucial, with all pulmonary blood flow originating from the aorta via a patent ductus arteriosus or major aortopulmonary collateral arteries.[3]

TOF with absent pulmonary valve (6% of TOF)

Although the pulmonary valve is absent, the right ventricular outflow tract remains open. Although these infants are typically acyanotic due to the absence of RVOTO, the condition is characterized by respiratory complications arising from massive aneurysmal dilation of the pulmonary arteries, which results from pulmonary valve absence and obligatory regurgitation.[4,6] These aneurysmal enlargements exert external pressure on the distal trachea and bronchi, leading to intrathoracic airway obstruction, lung atelectasis, and potentially pulmonary hypoplasia.[4]

Pathophysiology of TOF

The structural features of tetralogy of Fallot facilitate the blending of blood from the pulmonary and systemic circulations. Cyanosis results from deoxygenated blood entering the systemic circulation via a right-to-left shunt, a mixing process that typically takes place at the VSD. The direction of blood flow across the VSD depends on the pressure difference between the right and left ventricles. The severity of RVOTO dictates the volume of pulmonary blood flow, which corresponds to the right ventricular stroke volume.

RVOTO severity often involves both fixed anatomical and variable physiological factors.[3,5] Differences in these factors among individuals explain the varying saturation levels observed in these patients prior to repair. Grasping the dynamic elements that exacerbate or alleviate right-to-left shunting is essential for managing critically ill or pre-operative infants with TOF.

CASE REPORT

A 12 year female child was referred to us for clinical cardiac evaluation and transthoracic echocardiography (TTE). The child was full term normal delivery born out of consanguineous marriage. There was no history of maternal risk factors of congenital heart disease (obesity, diabetes, febrile illness, smoking, alcohol intake, teratogenic drug use , or radiation exposure. The history was narrated by the parents. They informed that the child was acyanotic since birth however they gave history of moderate to severe breathlessness while playing/climbing stairs/dancing. However, they denied any history of loss of consciousness, syncopal attacks (“Tet spells”), palpitations, or swelling over feet/face.

Clinical assessment revealed the patient had an average build and normal appearance (Fig. 1a). Although clubbing was observed (Fig., no cyanosis was detected through bluish discoloration of the lips, fingertips, toes, or nail beds (Fig 1a,1b,1c) The child weighed 39 kg, stood 149 cm tall, and exhibited a pulse of 69/min, blood pressure of 100/70 mmHg, respiratory rate of 15/min, and an SPO2 of 92% on room air. Peripheral pulses were all palpable and normal, with no radio-femoral delay. Cardiac assessment revealed a grade 4/6 systolic ejection murmur in the pulmonary region. The initial heart sound was normal, while the second was inaudible. Neither clicks nor gallop sounds were detected. The remainder of the systemic examination showed no significant findings.

The posteroanterior chest radiograph indicated slight cardiac enlargement accompanied by diminished pulmonary blood flow (Fig 1d).

The resting ECG showed normal sinus rhythm at 70 beats per minute, accompanied by right ventricular hypertrophy and right axis deviation (Fig. 1d).

Fig. 1a

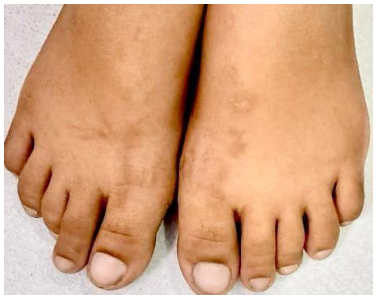

Fig. 1b

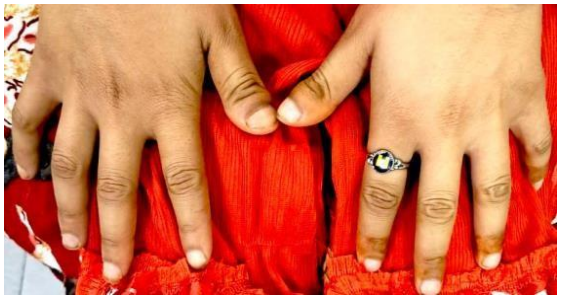

Fig.1c

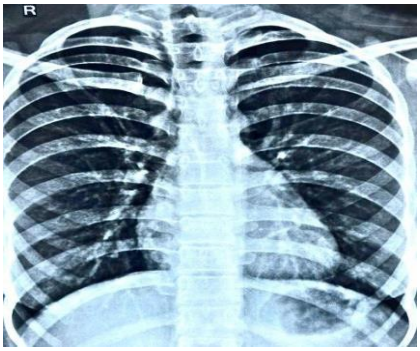

Fig. 1d

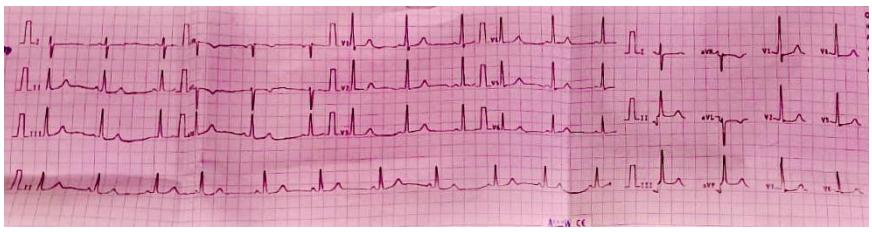

Fig. 1e

Figure 1: Clinical Features, X-ray chest & Resting ECG of our index patient. – a) Normal face, b) Clubbing of toes, c) Clubbing of fingers, d) X-ray chest (PA view), e) Resting ECG.

Transthoracic Echocardiography

The author conducted all echocardiographic assessments using an Esaote My Lab X7 4D XStrain system from Italy. Images were obtained with a pediatric probe featuring a harmonic variable frequency electronic single-crystal array transducer while the subject lay supine and in the left lateral decubitus position.

Standard echocardiographic assessments, including M-mode, 2D, and both pulse and continuous wave Doppler, were conducted using subcostal, parasternal long and short axis, four- and five-chamber, and suprasternal views. We performed a contemporary sequential segmental echocardiographic assessment on the index patient and listed the distinctive findings.

M-Mode Echocardiography.

M-mode echocardiography of the right and left ventricles was conducted, with the resulting estimates detailed in Table 1.

Table 1: Calculations of M-mode echocardiography.

Variables	LV	RV
IVS d	6.2 mm	- mm
LVID d	38.3 mm	26.0 mm
LVPW d	5.6 mm	8.6 mm
IVS s	8.0 mm	12.6 mm
LVID s	25.7 mm	15.3 mm
LVPW s	11.2 mm	9.9 mm
EF	62 %	74 %
% LVFS	33 %	41 %
LVEDV	63.1 ml	24.6 ml
LVESV	23.9 ml	6.3 ml
SV	39.2 ml	18.2 ml
LV Mass	58 g	42 g

Summary of M-mode echocardiography

Characteristic right ventricular hypertrophy was observed, with normal size and function in both ventricles. Biventricular ejection fractions were 62% and 74%, while the corresponding masses were 58g and 42g.

2-Dimensional Color Echocardiography

Transthoracic color echocardiography exhibited multiple features as mentioned below.

1. Levocardia (Fig.1d). Situs Solitus (Fig.2a). AV concordance.

VA concordance.

D-loop ventricles (Fig.2b).

D-loop great arteries (normally related great arteries) (Fig.2c). Left aortic arch. (Fig.2d).

Confluent pulmonary arteries.

Normal pulmonary and systemic venous drainage.

2. TETRALOGY OF FALLOT'S

a) VENTRICULAR SEPTAL DEFECT (Large)(Fig.2e)

- Size – 14.5mm.
- Subaortic, Malaligned, Perimembranous, non-membranous type
- Bidirectional shunt, Predominantly Lt to Rt. Shunt.

b) Overriding of Aorta = 30% (Fig.2f).

c) Infundibular obstruction (Severe) (Fig.2g). PV Normal (Fig 2h).

Peak/mean gradient across RVOT 742/39.2 mmHg. (Fig.2i,2j) Hypoplasia of distal MPA, LPA & RPA. (Fig. 2k)

Ao annulus d (29.3 mm). PV annulus d (22.30 mm).

MPA proximal d (26.30 mm). MPA distal d (10.30 mm).

RPA d (7.10 mm). LPA d (11.50 mm).

d) Dilated RV with concentric hypertrophy of RV. Normal biventricular systolic function.

Normal LVEF = 62 % (M-mode estimation) (Fig.2l)

LVEF = 59 % (Simpson's biplane method) (Fig.2m). RVEF = 74 % (Fig 2n).

3. No evidence of ASD, PDA, COA, AS.

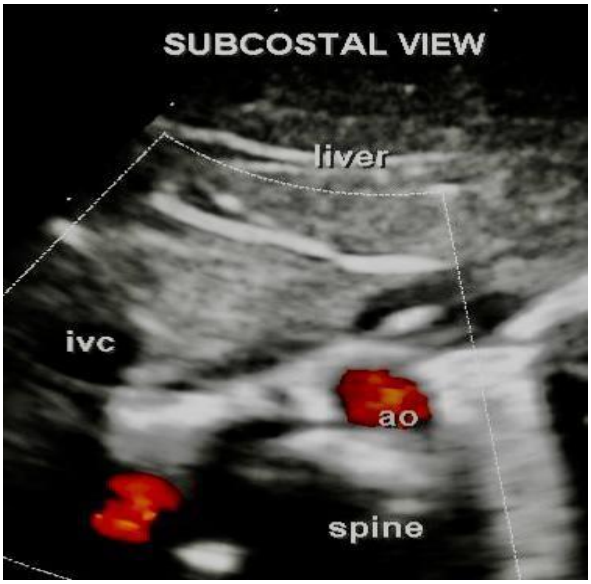

Fig. 2A

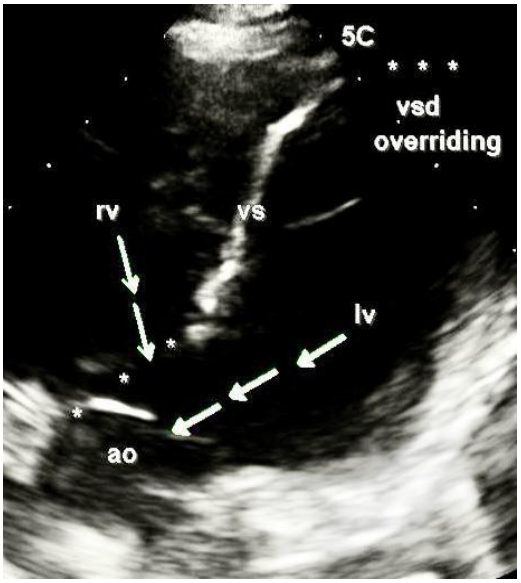

Fig. 2B

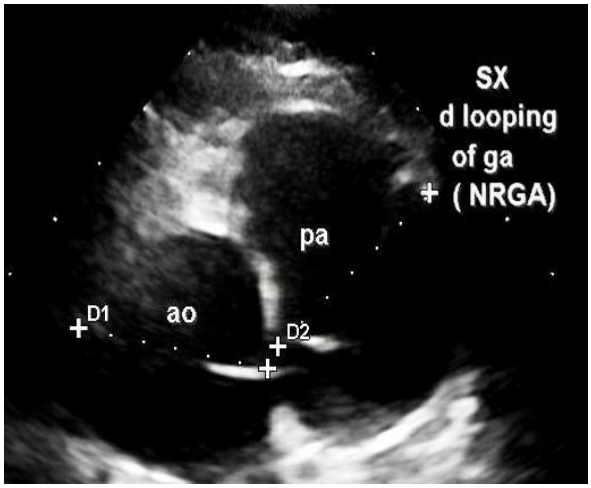

Fig. 2C

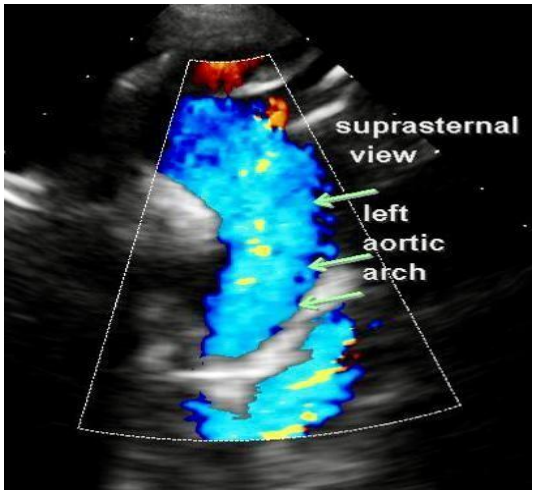

Fig. 2D

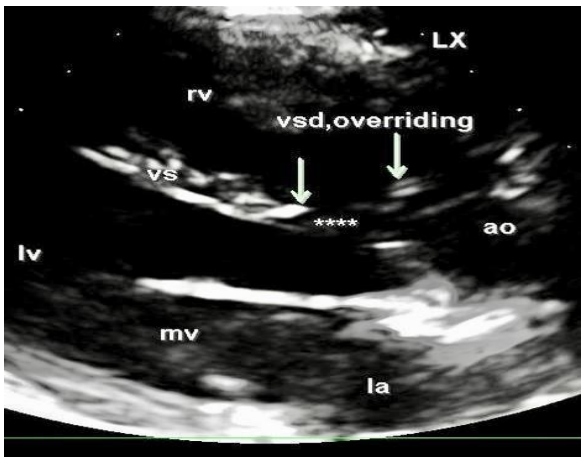

Fig. 2E

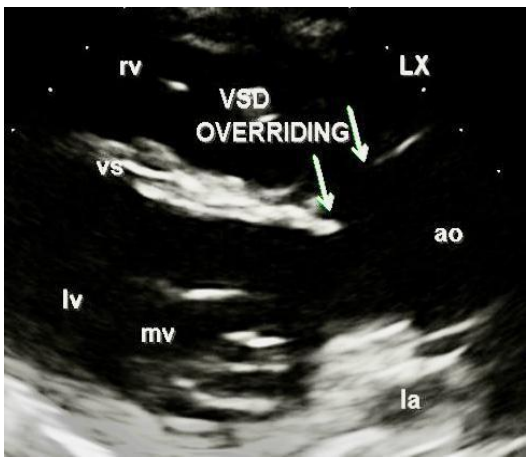

Fig. 2F

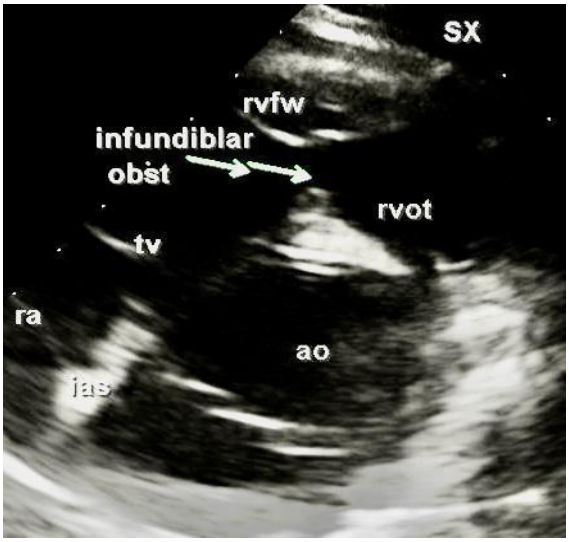

Fig. 2G

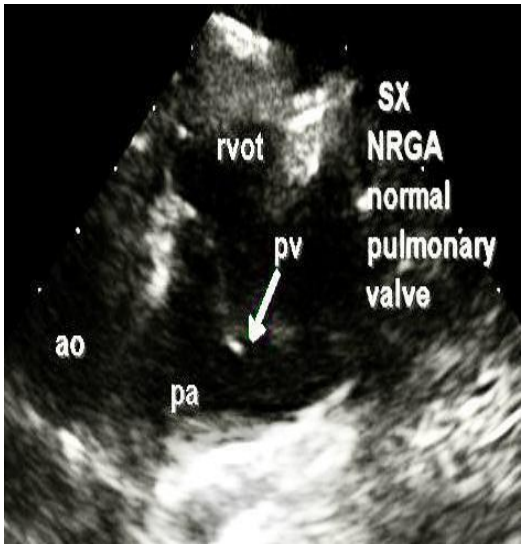

Fig. 2H

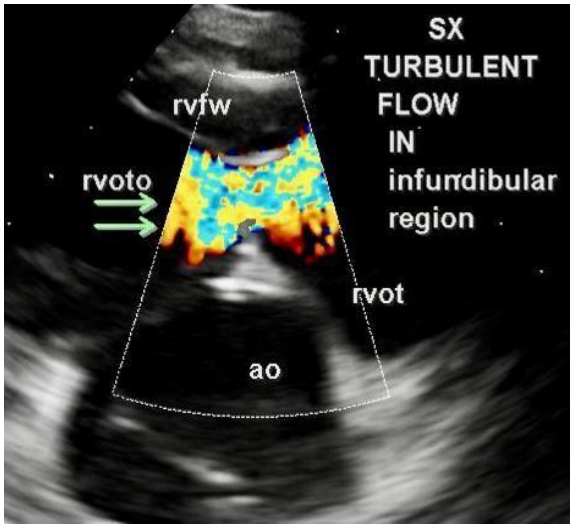

Fig. 2I

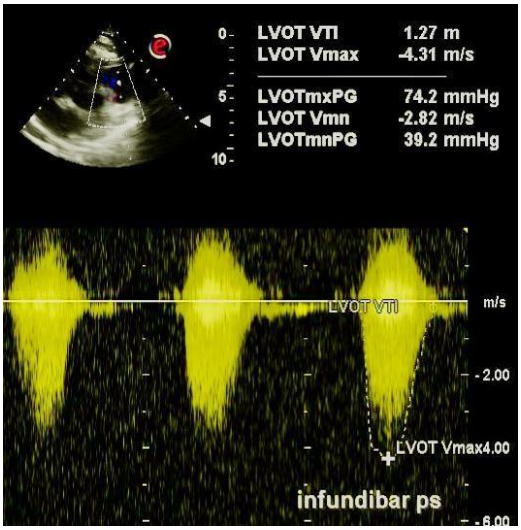

Fig. 2J

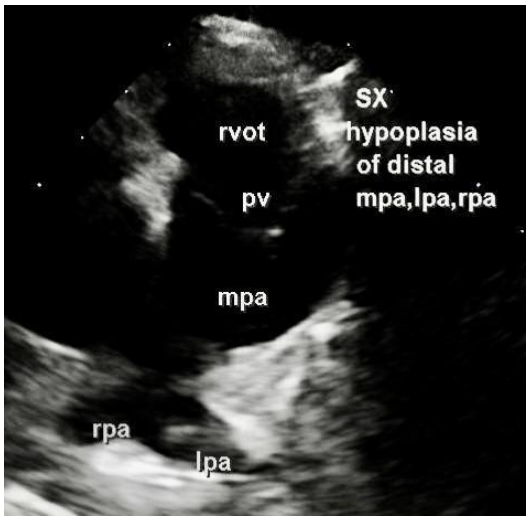

Fig. 2K

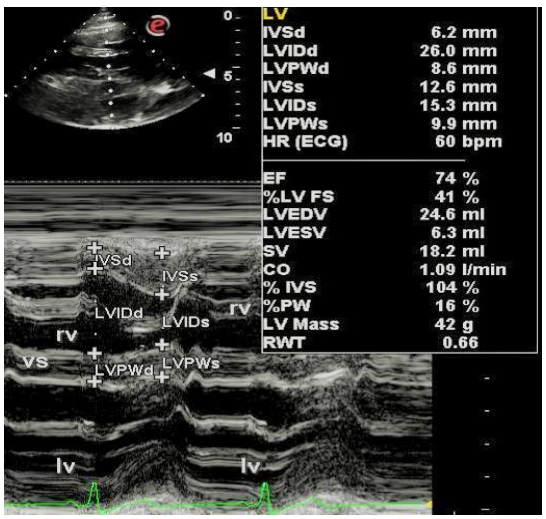

Fig.2L

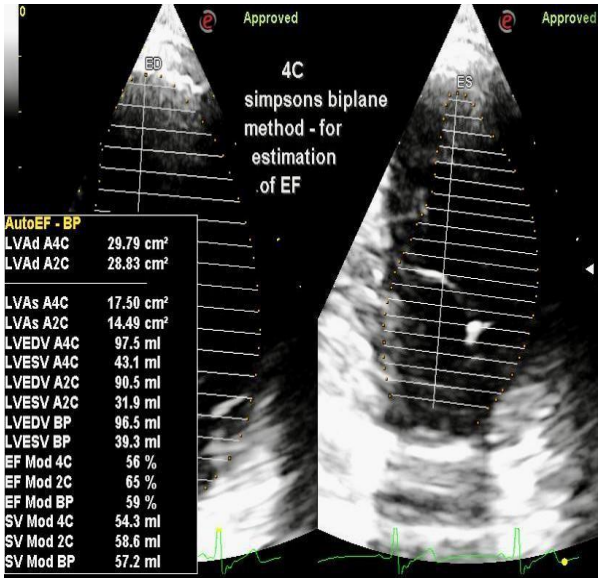

Fig. 2M

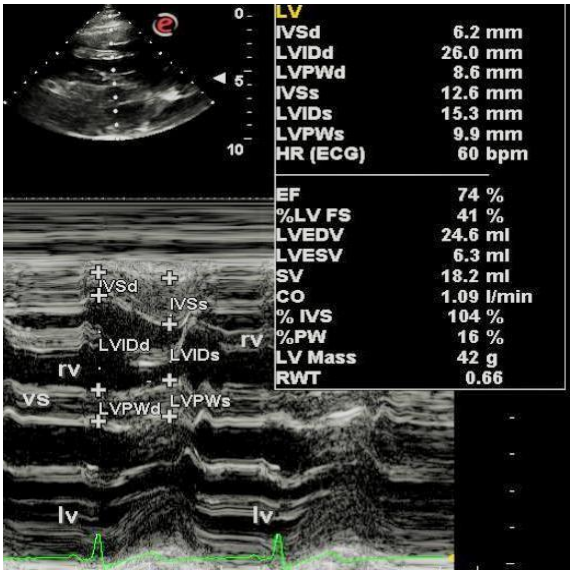

Fig. 2N

Figure 2: Transthoracic 2 Dimensional Color Echocardiographic Characteristics of our index patient. ; Fig. 2a, In the subcostal view normal situs solitus is visualised, with aorta (ao) on the left and inferior vena cava (ivc) on the right ; **Fig. 2b** , d looping of ventricles - right ventricle lying on the right side of left ventricle; **Fig. 2c** , d looping of great arteries (GA) ; **Fig. 2d** , left aortic arch ; **Fig. 2e**,VSD (Large) ; **Fig. 2f** , Overriding of aorta 30 % ; **Fig. 2g** , Infundibular obstruction- Turbulent mosaic flow pattern is demonstrated; **Fig. 2h** , normal pulmonary valve ; **Fig. 2i** , peak/mean gradient across infundibular region was 74.2/39.2 mmhg ; **Fig. 2k** , Hypoplasia of distal main pulmonary artery, left & right pulmonary arteries **Fig. 2l** , LVEF was 62 % derived by m- mode estimation; **Fig. 2m**, LVEF was 59 % by Simpson's biplane method ; **Fig. 2n** , RVEF was 74 % by m-mode method.

Summary of Transthoracic Color Echocardiography

Transthoracic color echocardiography demonstrated typical features of tetralogy of Fallot, despite the presence of mild aortic overriding (30%). The right ventricle exhibited dilation and concentric hypertrophy, while the left ventricle maintained normal cavity dimensions. Both ventricles exhibited normal function, with right and left ventricular ejection fractions of 72% and 59%, respectively.

Future course of action:

Our index patient, a 12-year-old girl with Pink Tetralogy of Fallot (92% room-air saturation), and moderate-to-severe exercise limitation was referred to a tertiary pediatric cardiovascular center for complete surgical repair.

DISCUSSION

Pink versus blue Tetralogy of Fallot

Pink tetralogy of Fallot describes cases with mild right ventricular outflow obstruction and slight cyanosis, whereas blue tetralogy denotes the classic form featuring severe obstruction, substantial right- to-left shunting, and pronounced cyanosis.

Pathophysiologic Distinction

The extent of right ventricular outflow tract obstruction, rather than the existence of a ventricular septal defect, distinguishes pink from blue TOF.[8]

Pink Tetralogy of Fallot.

- Even with a ventricular septal defect, mild pulmonary stenosis ensures sufficient blood flow to the lungs.[7]
- Insignificant or missing resting cyanosis results from inadequate RVOT obstruction to drive substantial right-to-left shunting.[9]
- The equilibrium between pulmonary stenosis and VSD stops deoxygenated blood from dominating the systemic circulation.[9]
- This condition is frequently mistaken for a straightforward VSD because of the prominent precordial murmur and the absence of clear cyanosis.[7]
- Diagnosis may be delayed until adulthood due to the subtle nature of the symptoms.[10]

Blue (Classic) Tetralogy of Fallot.

- Severe RVOT obstruction limits pulmonary blood flow.[8]
- Significant cyanosis usually emerges during the first year of life if it was not evident at birth.[9]
- Right-to-left shunting through the VSD delivers deoxygenated blood to the systemic circulation.[9]
- Clubbing develops in chronic cases.[7]
- To offset diminished pulmonary blood flow, patients can form numerous aorticopulmonary collateral vessels.[7]

Clinical Presentation Differences

Pink TOF physical Examination

- A loud systolic murmur at the precordium is frequently misdiagnosed as an isolated ventricular septal defect.[7]

- Normal or near –normal oxygen saturation.[7]
- Absence of clubbing.[7]
- A mild right ventricular outflow tract obstruction[7] may produce a soft ejection systolic murmur.

Blue TOF Physical Examination:

- Cyanosis and clubbing in unoperated patients.[7]
- The presence of aorticopulmonary collaterals results in loud, persistent murmurs across the chest.[7]
- Pulses may be reduced or missing on the side of a previous Blalock-Taussig shunt.[7]

Critical Diagnostic Pitfall

The primary clinical pitfall is failing to identify pink tetralogy of Fallout completely. Due to their mild cyanosis and prominent murmur, these patients are often incorrectly diagnosed with an isolated small VSD.[7]

The diagnosis may not be made until adulthood, delaying necessary surgical intervention.[10]

Key differences: Blue vs. Pink TOF.[11]

Feature	Blue TOF (Classic)	Pink TOF
RVOT Obstruction	Severe	MILD/Minimal
Cyanosis	Present (at rest or with activity)	Absent or minimal at rest
Shunt direction	Right-to-Left (Deoxygenated to Systemic)	Left-to-Right (Oxygenated to Pulmonary)
Pulmonary Flow	Decreased	Normal or increased
Presentation	“Blue Baby” hyper cyanotic spells	Often misdiagnosed as simple VSD,loud murmur
Symptom Onset	Birth to early infancy	Late childhood or adulthood
Physiology	Cyanotic CHD	Similar to large VSD

Detailed Differences

Pathophysiologic mechanism: In blue TOF, the severe pulmonary stenosis forces oxygen poor blood through the VSD into the aorta, causing cyanosis. In pink TOF, the obstruction is mild enough that pressure is lower, allowing blood to flow from left to right across the VSD, resulting in normal oxygenation, acting similar to a large isolated VSD.

Presentation: Pink TOF patients may present with pulmonary overcirculation symptoms (similar left-to- right shunt) rather than oxygen deficiency, such as rapid breathing, poor feeding and slow weight gain.

CONCLUSION

We reported a case of a 12-year-old girl with “Pink” Tetralogy of Fallot. The study suggested that children with primary congenital heart defects might not be diagnosed until late childhood, even with rapid medical progress. Postponing the diagnosis and treatment of TOF heightens the likelihood of unfavorable results. Comprehensive newborn physical exams and early-life echocardiographic screenings can facilitate the earlier detection of the disease.

Patients with "Pink" TOF are encountered very rarely. Because the pathophysiology of pink TOF differs significantly from that of blue TOF, this case represents a valuable contribution to the literature.

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