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Case Study

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TRANSTHORACIC COLOR DOPPLER ECHOCARDIOGRAPHY IN A CHILD AFFLICTED WITH "PINK" TETRALOGY OF FALLOT

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ABSTRACT
Tetralogy of Fallot (TOF) represents a prevalent congenital cardiac anomaly characterized by a constellation of four primary pathological features, alongside a spectrum of secondary morphological and functional anomalies. The cardinal components include an overriding aorta, a membranous ventricular septal defect (VSD), right ventricular hypertrophy, and right ventricular outflow tract (RVOTO) obstruction. The clinical prognosis of TOF is predominantly influenced by the degree of RVOTO, which modulates the degree of right-to-left shunting and subsequent systemic desaturation. Epidemiological studies indicate that survival beyond the fifth decade of life is possible, irrespective of surgical intervention, underscoring the variability in disease severity and management outcomes. Cyanosis, a hallmark of TOF, typically manifests within the first year of life if not present at birth. In cases where the RVOTO is mild and the VSD exhibits a balanced configuration, cyanotic episodes may be absent, a phenotype referred to as "Pink Tetralogy of Fallot." This presentation is associated with reduced right-to-left shunting due to preserved pulmonary blood flow. The present case describes a 12-year-old female of Indian origin diagnosed with Pink Tetralogy of Fallot, attributed to mild aortic overriding. A comprehensive echocardiographic assessment, including transthoracic color Doppler imaging, was conducted to delineate the anatomical and hemodynamic features of the condition.

KEYWORDS: Pink Tetralogy of Fallot (TOF), Overriding of Aorta, Right ventricular outflow obstruction, Tet spells, Echocardiography.

INTRODUCTION

TOF is a well-documented cyanotic congenital heart defect defined by four core anatomical abnormalities: a VSD, dynamic RVOTO, an overriding aorta, and right ventricular hypertrophy. The clinical expression and severity of the disorder are contingent upon the interplay of RVOTO severity, relative ventricular pressures, and the extent of aortic override relative to the VSD.**[Fig.1-4]**

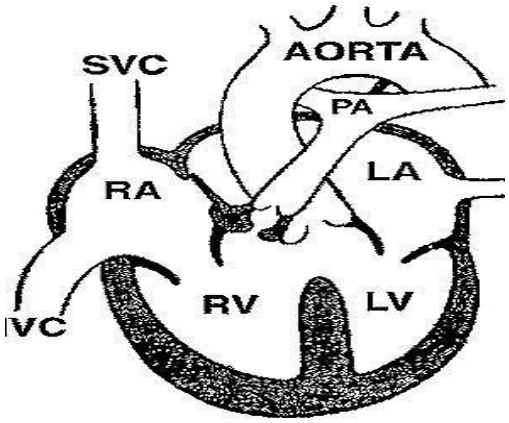


Fig. 1: This diagram depicts the features of Tetralogy of Fallot: 1. Ventricular septal defect; 2. Overriding aorta; 3. Pulmonic stenosis; 4. Right ventricular hypertrophy. The obstruction to right ventricular outflow creates a right-to-left shunt that leads to cyanosis.

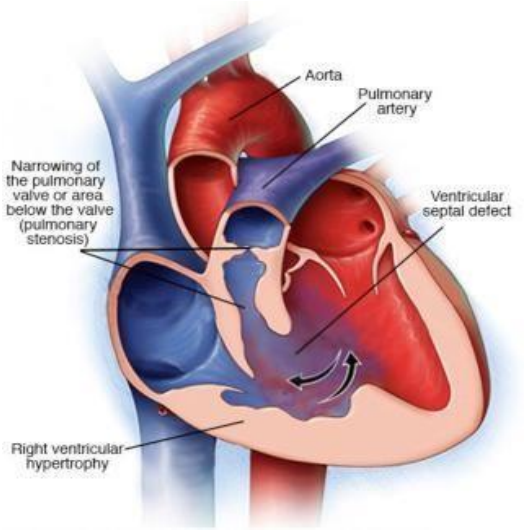


Fig. 2: Image demonstrating the classical TOF anatomy.

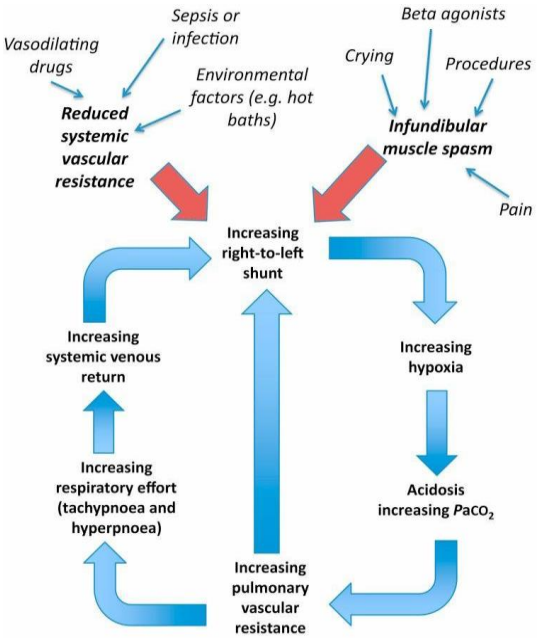
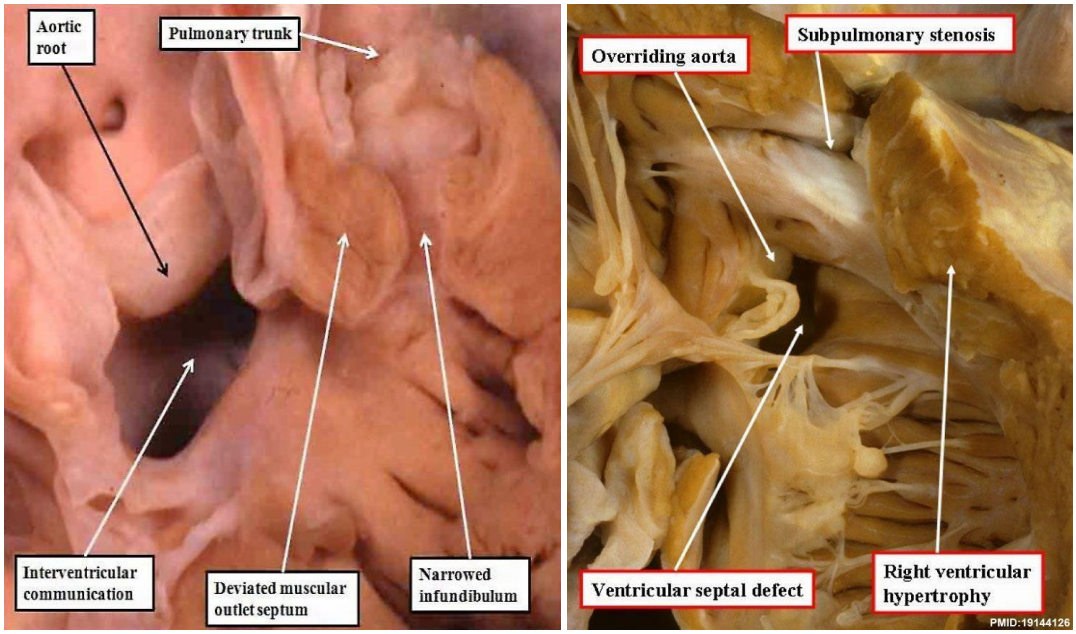


Fig 3: Mechanism and downward spiral of a cyanotic spell.



4a) 4b)

Fig. 4: Autopsy specimen showing features of Tetralogy of Fallot.

Genetic Considerations

Approximately 75–80% of TOF cases are classified as nonsyndromic, indicating the absence of associated systemic anomalies. Genetic studies have identified mutations in regulatory genes such as NOTCH1, FLT4, and TBX1, which are implicated in cardiac development and are frequently associated with 22q11.2 microdeletion syndromes.[1] Additional mutations in transcription factors critical to cardiac morphogenesis, including NKX2.5, GATA-6, GATA-4, HAND1, HAND2, ZFPM2, and NF-ATC, have also been linked to nonsyndromic TOF.[2,3] Familial recurrence of congenital heart defects (CHD) occurs in approximately 3% of affected families, though the specificity to TOF remains lower compared to the general population.[4]

In contrast, 20–25% of TOF patients exhibit syndromic associations or chromosomal abnormalities. The most common are Trisomy 21 (Down syndrome) and 22q11.2 microdeletion syndromes, which encompass a range of phenotypic expressions. The 22q11.2 deletion syndrome, for instance, may present with DiGeorge syndrome (characterized by dysmorphic facies, palatal anomalies, immune deficiencies, hypocalcemia, and learning disabilities) or velocardiofacial syndrome (lacking immune and calcium abnormalities). Notably, 22–48% of individuals with 22q11.2 microdeletions exhibit interrupted aortic arch, and 24% present with right-sided aortic arch, further complicating the clinical picture. The association between TOF and 22q11.2 microdeletion is particularly pronounced in cases of pulmonary valve atresia, where the prevalence of the deletion increases to 40%. Consequently, genetic testing for 22q11.2 microdeletion is increasingly recommended in prenatal diagnostics for TOF, given the adverse prognostic implications of this genetic variant.[5]

Pathophysiology of TOF

The pathophysiological mechanism of cyanosis in TOF arises from the blending of oxygenated and deoxygenated blood within the systemic circulation. This occurs via a right-to-left shunt at the VSD, driven by the pressure gradient between the right and left ventricles. The severity of RVOTO directly influences pulmonary blood flow, which in turn determines the volume of right ventricular output and the extent of shunting. RVOTO severity is multifactorial, involving both anatomical (e.g., pulmonary valve stenosis) and physiological (e.g., dynamic changes in ventricular pressures) components.[7,8] The variability in these factors among individuals accounts for the heterogeneous desaturation levels observed preoperatively. A nuanced understanding of the dynamic interplay between anatomical and physiological determinants of RVOTO is critical for the management of critically ill or pre-operative infants with TOF.[6]

Echocardiography in Tetralogy of Fallot[9]

Echocardiography is indispensable in the diagnostic and preoperative evaluation of TOF. It enables precise assessment of the VSD, including its location and size, the degree of aortic override, and the extent of RVOTO. Additional parameters evaluated include the severity of pulmonary stenosis, the morphology of the coronary and pulmonary arteries, and associated anomalies such as aortic arch abnormalities. The interventricular septum defect requires meticulous characterization, with approximately 80% of cases presenting as a perimembranous VSD, typically large and subaortic in location.

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 CHD (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. 5a).

Although clubbing was observed (Fig., no cyanosis was detected through bluish discoloration of the lips, fingertips, toes, or nail beds (Fig 5a,5b,5c)

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 5d).

The resting ECG showed normal sinus rhythm at 70 beats per minute, accompanied by right ventricular hypertrophy and right axis deviation (Fig. 5d).

Fig. 5a

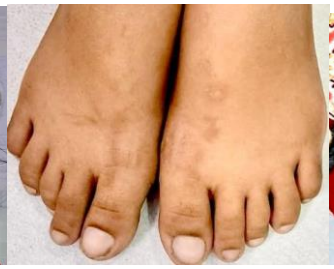


Fig. 5b

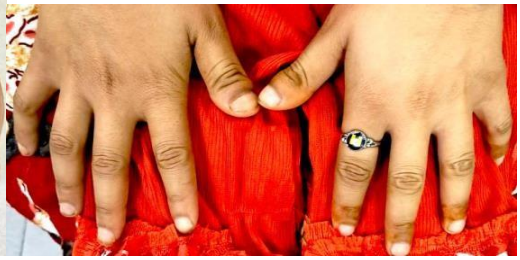


Fig.5c

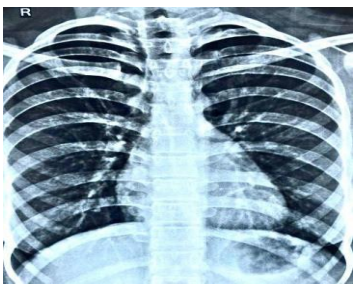


Fig. 5d

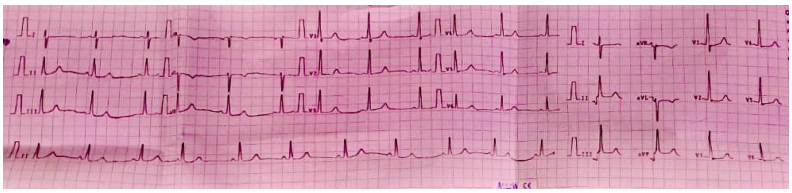


Fig. 5e

Figure 5: 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 perspectives. We performed a modern 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.
Levocardia (Fig.5d).
Situs Solitus (Fig.6a).
AV concordance.
VA concordance.
D-loop ventricles (Fig.6b).
left aortic arch (Fig. 6d)
D-loop great arteries. [Normally related great arteries (Fig. 6c)]
Confluent pulmonary arteries.
Normal pulmonary and systemic venous drainage.

1. TETRALOGY OF FALLOT'S
a) VENTRICULAR SEPTAL DEFECT (Large)(Fig.6e)
- Size – 14.5mm.
- Subaortic, Malaligned, Perimembranous, non-membranous type
- Bidirectional shunt, Predominantly Lt to Rt. Shunt.

b) Overriding of Aorta = 30% (Fig.6f).
c) Infundibular obstruction (Severe) (Fig.6g, Fig.6h, Fig.6i, Fig.6j).
Hypoplasia of distal MPA, LPA & RPA. (Fig. 6k)
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.6l)
LVEF = 59 % (Simpson's biplane method) (Fig.6m).
RVEF = 74 % (Fig 6n).

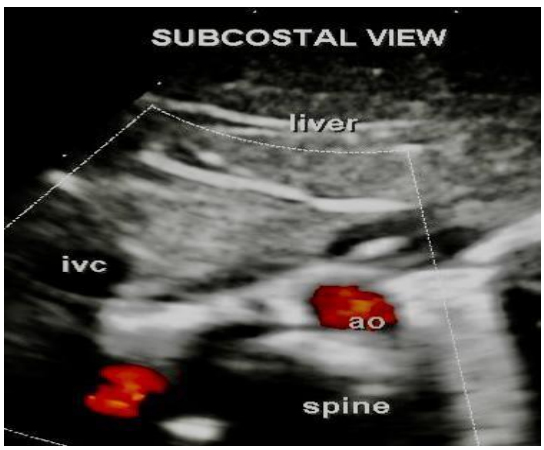


Fig.6a

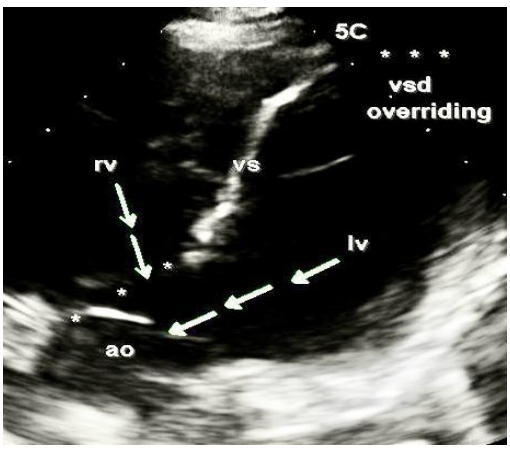


Fig.6b

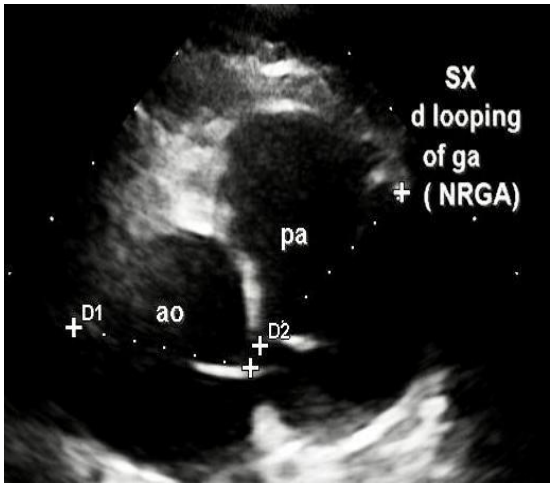


Fig.6c

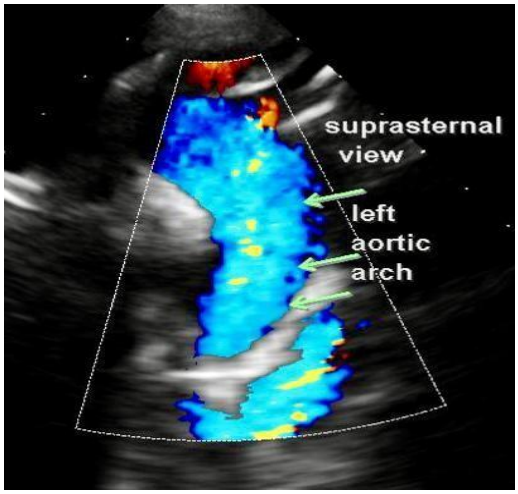


Fig.6d

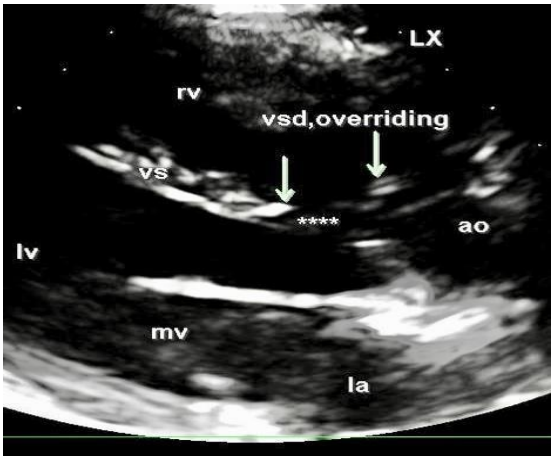


Fig.6e

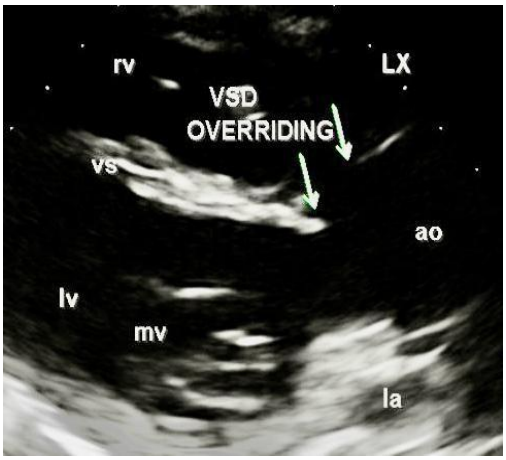


Fig.6f

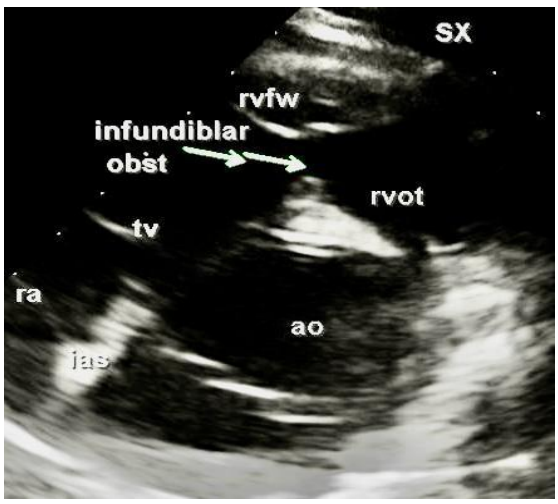


Fig.6g

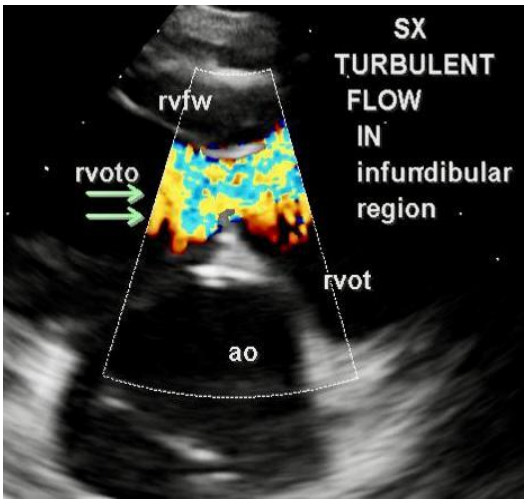
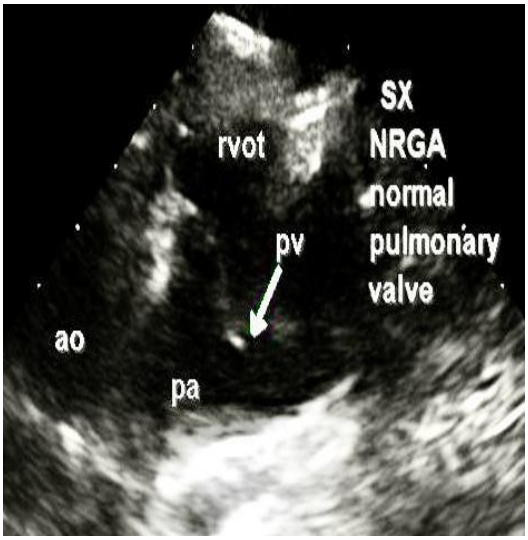


Fig. 6h



(Fig. 6i)

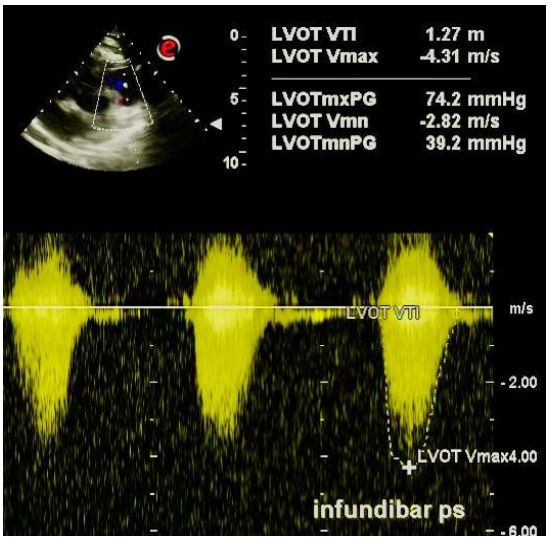


Fig.6j

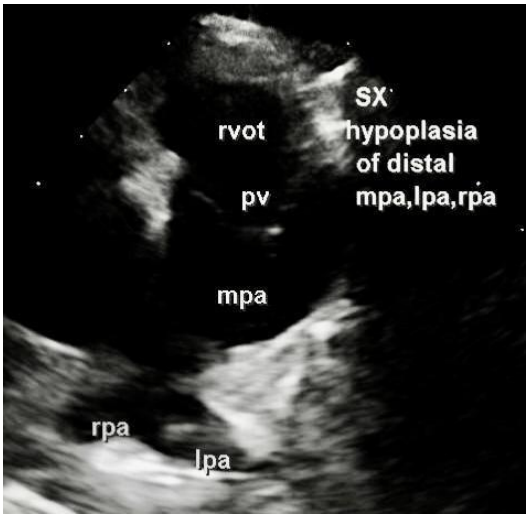


Fig.6k

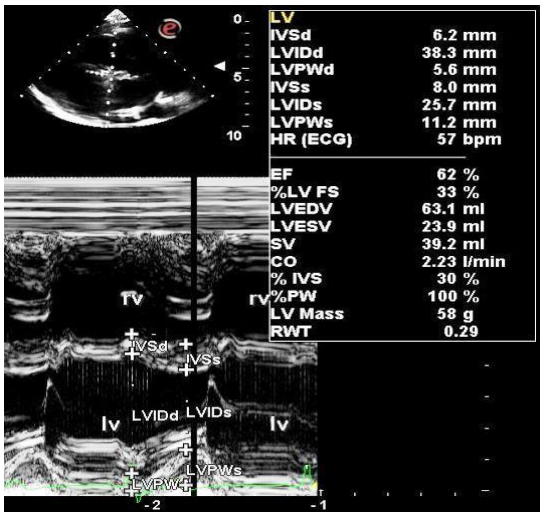


Fig.6l

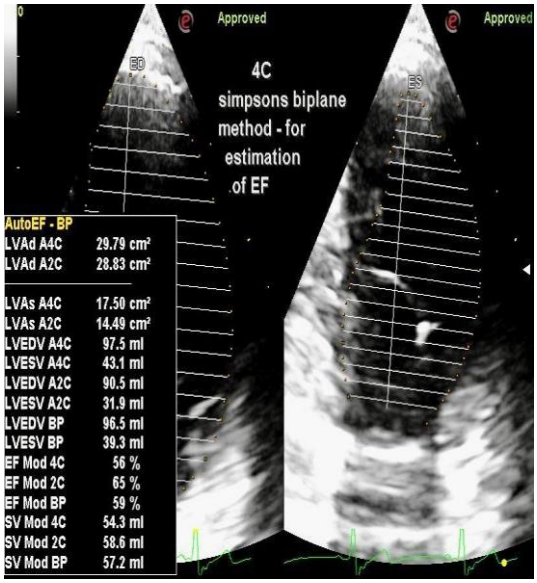


Fig.6m

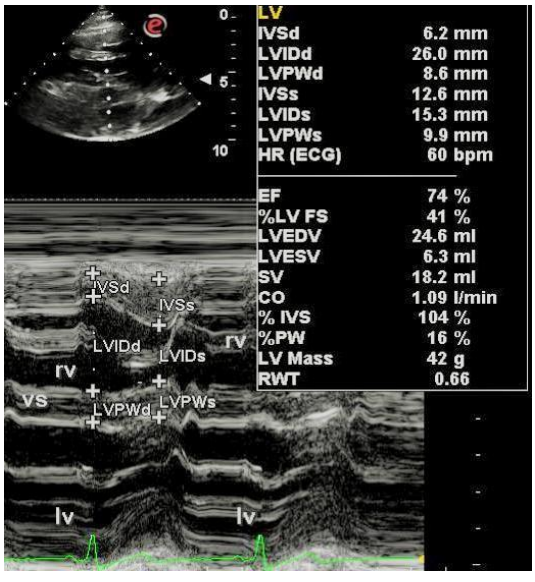


Fig.6n

Figure 6: Transthoracic 2 Dimensional Color Echocardiographic Characteristics of our index patient.; Fig. 6a, In the subcostal view normal situs solitus is visualised, with aorta (ao) on the left and inferior vena cava (ivc) on the right; Fig. 6b, d looping of ventricles - right ventricle lying on the right side of left ventricle; Fig. 6c, d

looping of great arteries (GA); Fig. 6d, left aortic arch; Fig. 6e,VSD (Large); Fig. 6f, Overriding of aorta 30%; Fig. 6g, Infundibular obstruction; Fig. 6h, Turbulent mosaic flow pattern is demonstrated; Fig. 6i, normal pulmonary valve; Fig. 6j, peak/mean gradient across infundibular region was 74.2/39.2 mmhg; Fig. 6k, Hypoplasia of distal main pulmonary artery, left & right pulmonary arteries Fig. 6l, LVEF was 62% derived by m- mode estimation; Fig. 6m, LVEF was 59% by Simpson's biplane method; Fig. 6n, 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

Table 2. Key differences: Blue vs. Pink TOF.[10]

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

Echocardiography in relation to TOF

Goals of Echocardiographic Exam[11-18]

Serial transthoracic echocardiography may yield critical diagnostic insights into the hemodynamic and anatomical characteristics of tetralogy of Fallot (TOF); however, the definitive classification of "pink" versus "blue" TOF remains a clinical judgment based on comprehensive hemodynamic and anatomical assessment. A systematic evaluation of cardiac anatomy should prioritize the following parameters:

- Morphological characteristics of the ventricular septal defect (VSD), including perimembranous, muscular, or doubly committed defects with anterior malalignment of the outlet septum (notably the presence of a fibrous remnant in doubly committed defects).
- Presence or absence of an atrioventricular septal defect (AVSD).
- Additional muscular VSDs.
- Right ventricular outflow tract (RVOT) obstruction, with specific attention to its anatomical location and degree of severity.
- Evaluation of dynamic subvalvar obstruction.
- Morphological and functional attributes of the pulmonary valve.
- Anatomical dimensions and morphological features of the main and branch pulmonary arteries.
- Subaortic and aortic valve morphology and functional status.
- Sources of pulmonary blood flow, including patent ductus arteriosus (PDA) and collateral pathways.
- Archal laterality and branching patterns of the great arteries.
- Anatomical configuration of the coronary arteries.
- Presence or absence of a persistent left superior vena cava (PLSVC), with or without a bridging vein.
- Assessment of thymic tissue, which may be associated with 22q11.2 deletion syndrome.

Detailed Analysis of Pulmonary Outflow Obstruction.

Dynamic RVOT obstruction is characterized by a late-peaking, "lobster claw" or dagger-shaped Doppler waveform, reflecting transient obstruction during systole. In contrast, fixed valvar or supravalvar obstruction typically presents with a mid-systolic peak and a more symmetric Doppler signal. Obstruction involving multiple levels (i.e., serial obstruction) may pose diagnostic challenges due to overlapping Doppler

patterns, which can result from the superimposition of fixed and dynamic components. The degree of obstruction is quantified based on hemodynamic and echocardiographic criteria, necessitating a nuanced interpretation of Doppler and anatomical data to distinguish between static and dynamic components.

Table 3. Degree of RVOT/PV obstruction.

Degree of obstruction	Doppler velocity	Gradient across RVOT/PV
Mild	< 3m/s	< 36 mmHg
Moderate	3-4 m/s	36-36 mmHg
Severe	> 4 m/s	> 64 mmHg

*Note dependence on pulmonary vascular resistance, presence of PDA, systemic blood pressure in presence of large VSD in unrepaired TOF.

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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