SVOA Paediatrics

ISSN: 2755-1660

ScienceVolks Open Access

Case Report

The Revelation of Spontaneous Correction of Malalignment Ventricular Septal Defect and Overriding of the Aorta in a Child with Pink Tetralogy of Fallot

Ashish Shrivastava MD[1]*, Snigdha Bhatia MD[2], Sanjeev Aggarwal MD[2], Raya Safa MD[2]

Abstract

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease. The presentation and treatment approach depends on the severity of the right ventricular outflow tract obstruction, and most cases require a surgical correction. We describe a neonate with pink TOF who exhibited typical echocardiographic characteristics such as aortic overriding, and a large VSD with minimal or no pulmonary outflow tract obstruction. The patient lost to follow-up, and a repeat 5-year echocardiogram revealed the absence of aortic overriding and significantly restricted VSD with a left-to-right shunt (pressure gradient ~50 mmHg across the VSD). There is no previous evidence of spontaneous correction of the defects associated with TOF which makes this case so intriguing.

Keywords: *Tetralogy of Fallot, Ventricular septal defect, Echocardiogram*

Introduction

Tetralogy of Fallot (TOF) is a cyanotic congenital heart disease with four components including a significant non-restricted ventricular septal defect (VSD), overriding of the aorta, right ventricular outflow tract obstruction, and right ventricular hypertrophy (1). It accounts for 7 to 10 percent of all congenital heart defects. The etiology of the development of TOF in intrauterine life is multifactorial. Some of the risk factors include uncontrolled maternal diabetes, maternal exposure to retinoic acid, phenylketonuria, trisomy 21, trisomy 18, trisomy 13, and other chromosomal anomalies (7). Out of the 4 lesions, the degree of the right ventricular outflow tract obstruction (RVOTO) usually determines the degree of cyanosis and the severity of the symptoms (1). Depending on the degree of RVOTO it can be classified as 1) Pink TOF- with minimal or no RVOTO, 2) Blue TOF- classic with a degree of RVOTO, 3) Profound cyanosis TOF- severe or complete RVOTO.

The majority of the cases are diagnosed antenatally by fetal echocardiogram. An infant born with TOF may show a 'Boot Shaped" heart on a chest X-ray. Electrocardiogram depicts right axis deviation (RAD) due to underlying right ventricular hypertrophy (RVH). Cardiac magnetic resonance imaging and catheterization can be useful but not routinely used (7). Most of the defects tend to persist including a VSD with TOF (8). The standard of care depends on the severity of the presentation. Severe RVOTO usually requires prostaglandin infusion to keep the ductus arteriosus open. Surgical corrections are usually late and involve a patch closure of the VSD, and repair of the RVOTO if any (9, 11).

[1]The Heart Institute, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio 45229, USA

[2]Division of Pediatric Cardiology, Department of Pediatrics, Children's Hospital of Michigan, Detroit, MI, USA.

***Corresponding Author:** Ashish Shrivastava, Department of Pediatric Cardiology, Cincinnati Children's Hospital Medical Center 3333 Burnet Ave, Cincinnati, Ohio-45220, USA.

https://doi.org/10.58624/SVOAPD.2025.04.003

Received: February 03, 2025

Published: February 22, 2025

Citation: Shrivastava A, Bhatia S, Aggarwal S, Safa R. The Revelation of Spontaneous Correction of Malalignment Ventricular Septal Defect and Overriding of the Aorta in a Child with Pink Tetralogy of Fallot. *SVOA Paediatrics 2025,* 4:1, 13-16. doi: 10.58624SVOAPD.2025.04.003

Case Presentation

Our patient was born at 34 weeks of gestation by C-section, Twin A, female, of a monochorionic diamniotic pregnancy. Her birth weight was 1445 grams (small for gestational age). APGARs at birth were 8 and 9 at 1 and 5 minutes respectively and saturations at birth were 98-99%. There were no complications during pregnancy or her birth. An echocardiogram (ECHO) performed at birth was consistent with the diagnosis of pink TOF including large unrestricted outlet VSD and overriding aorta (Figures 1). She was transferred to the neonatal intensive care unit (NICU) for further care.

There was no evidence of RVOT obstruction and had normal-sized confluent pulmonary arteries. She stayed in the NICU for 2 weeks for management of prematurity and hyperbilirubinemia and did not require any respiratory support. She did not develop any signs of pulmonary over-circulation and saturations remained in the 98-99% range at the time of discharge. The follow-up ECHO before discharge shows no changes from the previous ECHOs. She was discharged in a stable condition with plans to follow up in the outpatient pediatric cardiology clinic.

After discharge from NICU, she was lost to follow-up and presented again at 5 years of age as a referral from her primary care physician for evaluation of a murmur. Her clinical presentation was not suggestive of cardiac symptoms including cyanosis, respiratory distress, or failure to thrive. She was able to keep up with her peers and was growing appropriately for her age (weight 18.8 kg at the 50th percentile). Her physical exam was significant for an III-IV/VI pan systolic murmur best heard over the left lower sternal border. An echocardiogram demonstrated significantly restricted small VSD with a small left to right shunt (Figures 2 and 3). The peak gradient across the VSD was ~50 mm Hg There was no evidence of right ventricular outflow obstruction. She was discharged from the clinic with plans to follow-ups with a pediatric cardiologist.

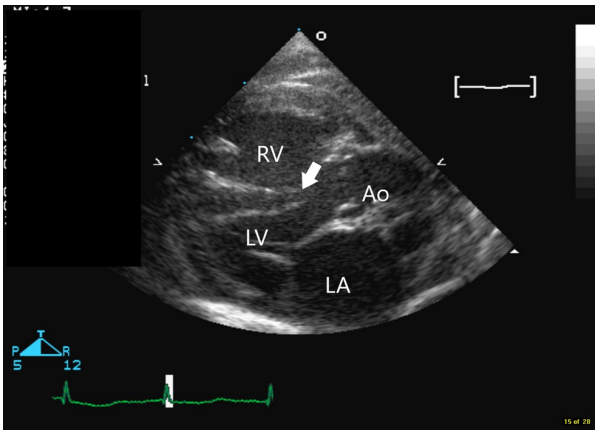

Figure 1. *Parasternal long-axis view showing ventricular septum with the overriding aorta. The arrow shows the site of VSD. RV: right ventricle, Ao: ascending aorta, LV left ventricle, and LA: Left atrium.*

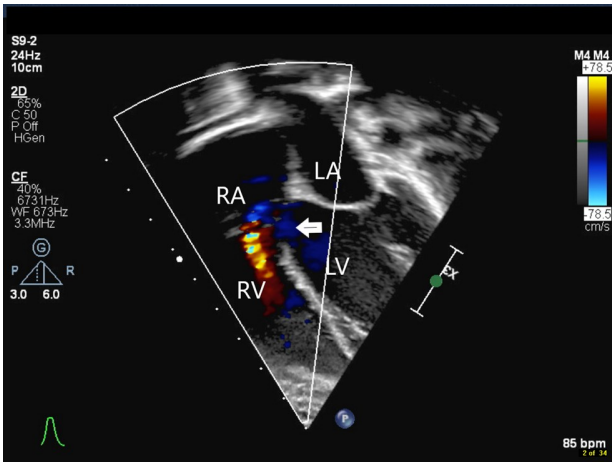

Figure 2. *Apical four-chamber view showing restricted VSD with the left to right shunt on color Doppler. The arrow shows the site of VSD. RV: right ventricle, RA: right atrium, LV left ventricle, LA left* atrium.

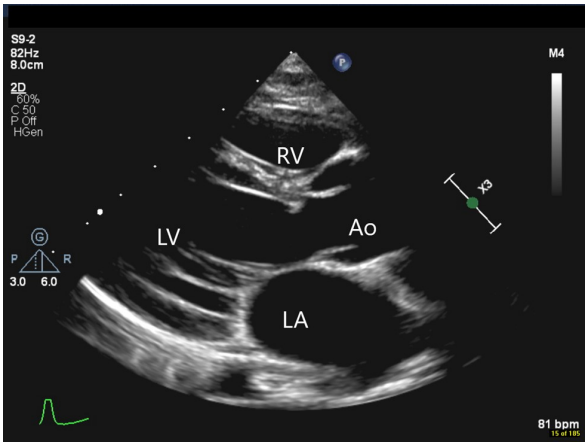

Figure 3. *Parasternal long-axis view showing no significant defect on 2D compared to a previously visualized malalignment in Image 1 at 5 years of life. RV: right ventricle, Ao: ascending aorta, LV left ventricle, and LA: Left atrium.*

Discussion and Conclusions

Michael EA et al. mentioned the evidence of a higher rate of re-intervention with an early age repair. Hence, most centers perform palliative repair later in the age of the patient is stable(10). Spontaneous closure of an isolated VSD is not uncommon and depends upon the location and size of the VSD. Small to moderate size perimembranous and muscular VSD tend to close spontaneously in the first year (mean (SD) 7.31 ± 6 months) and muscular VSD carries the highest rate of spontaneous closure (2). The probability of closure of VSD is highest in the first 6 months of life and depends on the location of the VSD rather than the size (3). Membranous VSDs typically close by adherence of tricuspid valve leaflet to the margin of the defect, aneurysm of the membranous septum, or prolapse of aortic valve leaflet through the defect (4). On the other hand, muscular VSDs are known to close secondary to growth and hypertrophy of the surrounding muscular septum and apposition of the defect by fibrous tissue.

The TOF kind VSD is large and spontaneous closure is rarely reported. There is no reported case of spontaneous closure or a significant decrease in the size of anterior malaligned TOF –type of VSD. Our case had large anterior malaligned VSD with the overriding aorta and became significantly small with no hemodynamic concerns. Wu et al described the spontaneous closure of anterior septal malalignment with isolated VSD in a cohort of 68 patients from China. They reported most of these VSDs became small due to aortic valve prolapse and aneurysm formation (5). On the other hand, our patient did not have any aortic valve changes associated with the decrease in the size of the VSD. Marino et al reported closure of a possible VSD in a patient with a double outlet right ventricle and pulmonary stenosis due to malalignment of the infundibular septum. Examination of the patient's heart on autopsy did not show a VSD, but there was indirect evidence of left ventricular dilation and hypertrophy suggestive of a previously present VSD. They hypothesized the progressive growth of accessory fibrous tissue from the aortic cusp to the muscular septum may have resulted in the closure of VSD (6).

TOF patients usually develop worsening cyanosis over time due to worsening RVOT obstruction and surgical repair remains the mainstay of management. On the contrary, the patient under discussion remained asymptomatic due to minimal RVOT obstruction. This is a unique case where malalignment ventricular septal defect with overriding of the ventricular septum by the aortic root spontaneously reduced to only a small perimembranous VSD with no hemodynamic concerns.

Conflict of Interest

The authors declare that they have no conflict of interest.

Funding

No funding was obtained for this study.

Ethical approval

This article does not contain any studies with human participants or animals performed by any of the authors.

Informed consent

Informed consent was obtained from the legal guardian of the patient included in the case report.

References

1. Karl TR, Stocker C. Tetralogy of Fallot and its variants. Pediatr Crit Care Med. 2016;17(Suppl. 1): S330–S336. https://doi.org/10.1097/pcc.0000000000000831
2. Li, X, Song, GX, Wu, LJ et al. Prediction of spontaneous closure of isolated ventricular septal defects in utero and post-natal life. BMC Pediatr 16, 207 (2016). https://doi.org/10.1186/s12887-016-0735-2
3. Hiraishi S, Agata Y, Nowatari M et al. Incidence and natural course of trabecular ventricular septal defect: two-dimensional echocardiography and color Doppler flow imaging study. J Pediatr. 1992;120(3):409–415. https://doi.org/10.1016/s0022-3476(05)80906-0

4. Zhang J, Ko JM, Guileyardo JM et al. A review of spontaneous closure of ventricular septal defect. Proc (Bayl Univ Med Cent). 2015; 28(4):516-520. https://doi.org/10.1080/08998280.2015.11929329

5. Wu MH, Wang JK, Chang CI, et al. Implication of anterior septal malalignment in isolated ventricular septal defect. Br Heart J 1995; 74:180-185. http://dx.doi.org/10.1136/hrt.74.2.180

6. Marino B, Loperfido , Savino Sardi C. Spontaneous closure of ventricular septal defect in a case of double outlet right ventricle. Br Heart J 1983; 49:608-11. https://doi.org/10.1136/hrt.49.6.608

7. Diaz-Frias J, Guillaume M. Tetralogy of Fallot. [Updated 2022 Jan 18]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK513288/

8. Wilson R, Ross O, Griksaitis MJ. Tetralogy of Fallot. BJA Educ. 2019 Nov;19(11):362-369. doi: 10.1016/j.bjae.2019.07.003. Epub 2019 Oct 14. PMID: 33456859; PMCID: PMC7807827

9. Rahmath MRK, Boudjemline Y. Tetralogy of Fallot Will be Treated Interventionally Within Two Decades. Pediatr Cardiol. 2020 Mar;41(3):539-545. doi: 10.1007/s00246-020-02297-z. Epub 2020 Mar 20. PMID: 32198584; PMCID: PMC7170827

10. Michael EA, Cunningham MD, Donofrio MT. Optimal timing for elective early primary repair of tetralogy of Fallot: analysis of intermediate term outcomes. *Ann Thorac Surg.* 2017;103:845–852

11. Bailliard F, Anderson RH. Tetralogy of Fallot. Orphanet J Rare Dis. 2009 Jan 13;4:2. doi: 10.1186/1750-1172-4-2. PMID: 19144126; PMCID: PMC2651859.