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An Unusual Case of A Neonate With Tetralogy of Fallot Physiology With Signs of Congestive Cardiac Failure

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ABSTRACT

Tetralogy of Fallot physiology is a unique pathophysiology centered around the interplay of pulmonary and systemic blood flow. Pulmonary atresia with an intact ventricular septum (PA-IVS) is a rare congenital heart defect characterized by a complete blockage of the pulmonary valve, preventing blood flow from the right ventricle to the pulmonary arteries. When PA-IVS occurs with TOF physiology, it results in a unique and unusual presentation due to the overlapping physiological effects of both conditions. Typically, in PA-IVS, the right ventricle (RV) is underdeveloped and unable to handle the pressure required for pulmonary circulation. However, in the presence of TOF physiology, the overriding aorta and the VSD may allow some oxygenated blood to reach the body, often leading to cyanosis. In this case, however, there was no VSD or overriding aorta. The finding of pulmonary regurgitation (PR) in this case is particularly unusual. PR occurs when blood flows backward into the right ventricle from the pulmonary artery, which is typically seen after surgical repair of TOF, especially when a patch is used to close the VSD or if residual pulmonary stenosis persists.The presence of PR in this patient may reflect an anomalous development of the pulmonary circulation, especially given the complex anatomy of PA-IVS and TOF physiology.

Keywords: *Tetralogy of Fallot (TOF), Pulmonary Atresia with Intact Ventricular Septum (PA-IVS), Congenital Heart Defect, Pulmonary Regurgitation (PR), Cyanotic Heart Disease, Right Ventricular Hypoplasia*

1. INTRODUCTION

Tetralogy of Fallot (TOF), one of the most common cyanotic congenital heart defects, is defined by four primary components: right ventricular outflow tract obstruction, a ventricular septal defect, an overriding aorta, and right ventricular hypertrophy. [1] The first is a VSD, which permits a shunt from right to left. The second is pulmonary circuit blood flow restriction caused by RVOT occlusion. It should be noted that in these patients, a right-to-left shunt across the VSD results from the RV's pressure being higher than the left ventricle's due to RVOT obstruction.[2]. The patient's heart auscultation revealed a systolic ejection murmur that is compatible with either pulmonic valvular stenosis or RVOT obstruction. [3] More deoxygenated and

oxygenated blood can enter the systemic circulation through the third, an overriding aorta. Lastly, the additional effort required to pump blood through the RV obstruction and overriding aorta causes RV hypertrophy. In this instance, the patient's chest radiograph and ECG match the RV enlargement observed in TOF. The degree of cyanosis and the time of onset are frequently correlated with the degree of right ventricular outflow tract blockage (RVOTO). In patients with TOF, right ventricular failure is rare but can happen under certain conditions.[4] The most prevalent genetic heart condition, hypertrophic obstructive cardiomyopathy (HOCM), affects at least 1 in 500 people in the general population. Because of mutations in 11 or more sarcomere or nearby Z-disc genes, it exhibits an autosomal dominant inheritance. Genetic testing distinguishes HOCM from other conditions that mirror it because of left ventricular hypertrophy, including Fabry's disease, Lysosome-associated membrane protein 2, and protein kinase AMP-activated non-catalytic subunit gamma 2.[5] Very few occurrences of TOF and HOCM together have been documented in the literature, making this combination exceedingly uncommon. Typically, pulmonary stenosis in TOF leads to reduced pulmonary blood flow, resulting in cyanosis. However, in rare cases, TOF physiology can be complicated by pulmonary atresia, with PR commonly seen postoperatively adding to the complexity. PR in a patient with PA-IVS and TOF physiology is highly unusual. This case explores the clinical presentation, diagnostic evaluation, and management challenges associated with this rare congenital heart defect.

2. CASE REPORT

A full-term male neonate, born via caesarean section due to a previous caesarean, was admitted to the NICU with respiratory distress and cyanosis shortly after birth. The baby, delivered to a 28-year-old mother (G2P1L1) at 38+6 weeks of gestation, showed no maternal comorbidities or history of teratogen exposure during pregnancy. A Fetal echocardiogram performed at 32 weeks revealed a dilated right ventricle, pulmonary artery dilatation, and a thickened dysplastic pulmonary valve. Upon NICU admission, the neonate showed no dysmorphic features or external congenital anomalies. Cyanosis persisted, with oxygen saturation between 75-80% on room air. All peripheral pulses were palpable without radio-femoral delay, and blood pressure was normal in all four limbs. Physical examination showed a weight of 3.5 kg, length of 48 cm, and head circumference of 39 cm, all within the 50th percentile. A grade 3/6 systolic murmur was heard along the left sternal border. but no signs of cardiac failure. Initial chest X-ray showed oligemic lung fields with cardiomegaly with CT ratio >0.6 Despite supplemental oxygen, the neonate remained hypoxemic, raising suspicion of a significant cardiac defect. A 2D echocardiogram performed on the first day of life revealed a moderate PDA with left-to-right shunting, a doming pulmonary valve with severe pulmonary stenosis (PS) and PR, mild mitral regurgitation (MR), and mild tricuspid regurgitation (TR). These findings suggested PA-IVS physiology. By day five, the neonate developed worsened respiratory distress signs of congestive cardiac failure and feeding difficulty. The patient was put on High flow nasal cannula support and diuresis was commenced with an injection of furosemide. Neonate responded to this treatment in the next three days. Repeat 2d-echo showed congenital valvular moderate pulmonary stenosis with significant pulmonary regurgitation small PDA with a left to right shunt small PFO with a left to right shunt dilated right ventricle intact IVS good LV and RV function. At the time of discharge patient was breastfed was having weight gain and was on diuresis. On 2 consequent follow-up patient has achieved the first milestone of social smile.

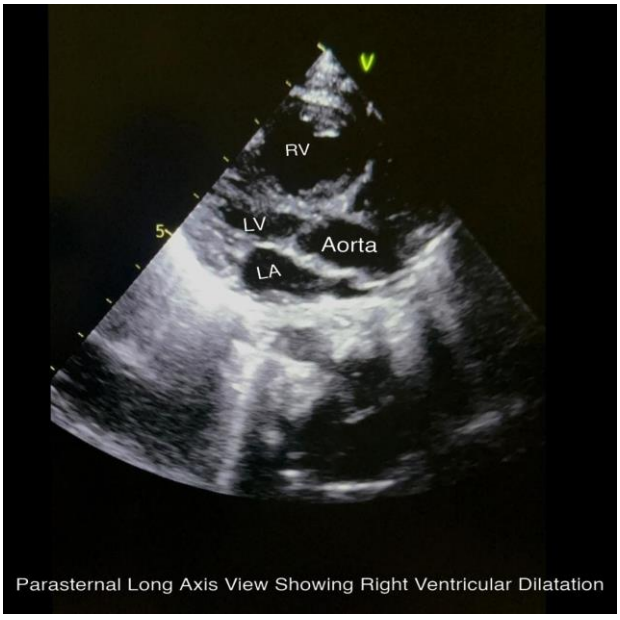

Figure 1: shows Parasternal long axis view showing right ventricular dilation

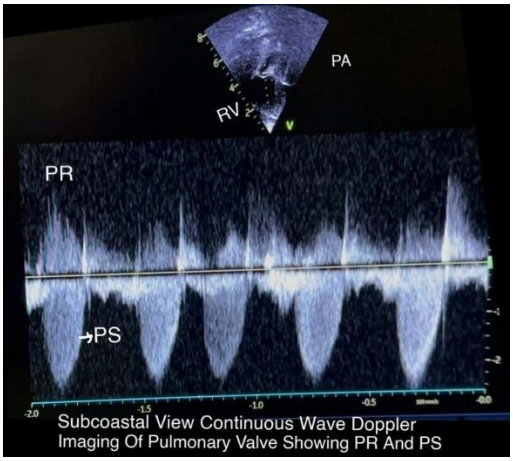

Figure 2: shows Subcoastal view continuous wave doppler imaging of pulmonary valve showing PR and PS

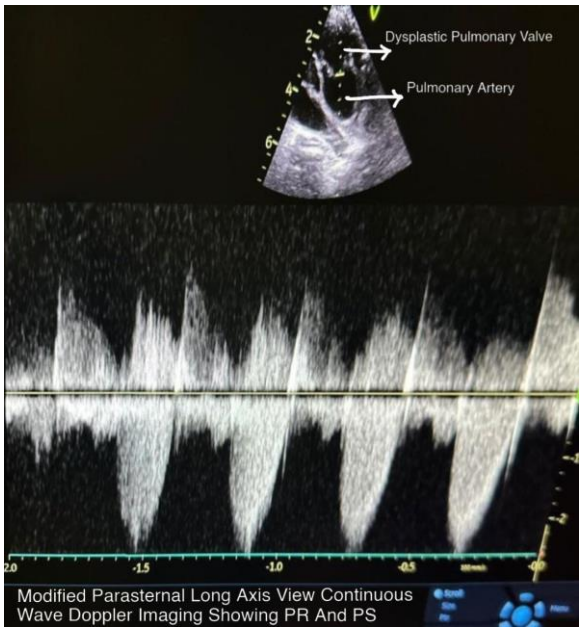

Figure 3: Modified parasternal long axis view continuous wave doppler imaging showing PR and PS

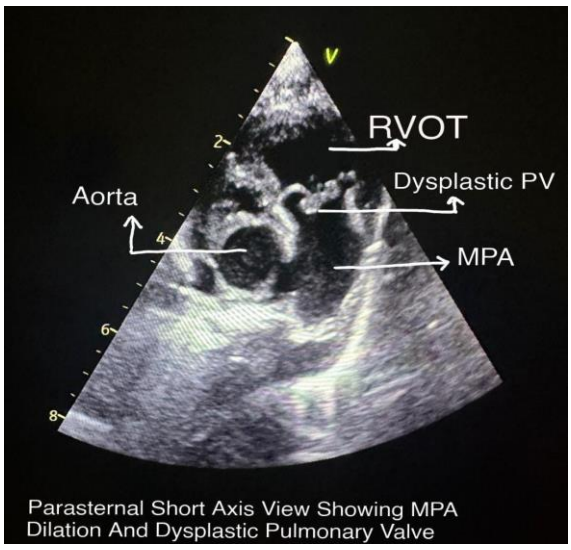

Figure 4: showing parasternal short axis view showing MPA Dilation and dysplastic pulmonary valve

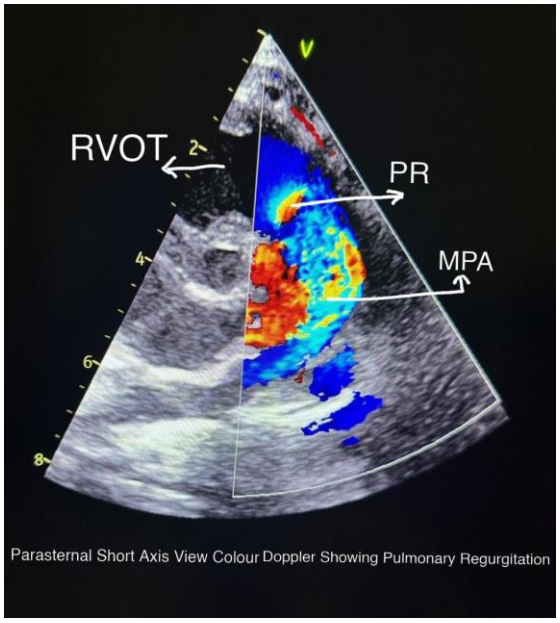

Figure 5: Parasternal short axis view colour doppler showing pulmonary regurgitation

3. DISCUSSION

The first case is that of a neonate presenting with pulmonary atresia and pulmonary stenosis along with intact ventricular septum with pulmonary regurgitation who presented with signs of congestive cardiac failure in the first week of life. Normally Tetralogy of Fallot physiology doesn't go into congestive cardiac failure due to pulmonary stenosis, right-to-left shunting, and reduced pulmonary blood flow it spares the heart from volume overload and prevents CCF in most cases. But in our case, the baby went into CCF due to pulmonary regurgitation. We have managed this by giving o2 support to high-flow nasal cannula and diuretics. The rarity of pulmonary atresia with intact ventricular septum, when it appears with signs of congestive cardiac failure, underscores the importance of thorough clinical evaluation. Neonates with this condition often present with cyanosis due to right-to-left shunting, but the addition of pulmonary regurgitation can exacerbate symptoms and lead to respiratory distress and heart failure [1]. The causes of congestive cardiac failure are Volume overload with cyanosis In our case on day 5 of life, the baby had severe respiratory distress and was on oxygen support and diuresis was started. Pulmonary atresia with intact ventricular septum (PA-IVS) is a rare and complex congenital heart defect that presents significant challenges in both diagnosis and management. This condition is marked by the absence of a functional connection between the right ventricle (RV) and the pulmonary artery, necessitating alternative pathways for pulmonary blood flow, such as a patent ductus arteriosus (PDA) or systemic-to-pulmonary collateral vessels (MAPCAs) (1,2).In most cases, pulmonary regurgitation (PR) is observed in postoperative settings, particularly after surgical repairs involving right ventricular outflow tract reconstruction or the placement of transannular patches[6]. Its occurrence in unoperated cases, is highly unusual which is present in our case and suggests an anomalous pulmonary vasculature or abnormal flow dynamics resulting from the associated defects [7]. Management strategies must be tailored to address both the immediate respiratory needs and the long-term implications for cardiac function. Early recognition and intervention are crucial, as delayed treatment can lead to significant morbidity and mortality. This case highlights the need for heightened awareness among clinicians regarding the potential presentations and complications associated with pulmonary atresia with intact ventricular septum, particularly in the neonatal population [8]. The coexistence of PA-IVS with Tetralogy of Fallot (TOF)-like physiology further complicates the clinical picture. Cyanosis in PA-IVS results from right-to-left shunting due to the inability of the RV to handle pulmonary circulation. [9,10] In this case, the addition of PR exacerbated the volume overload of the RV, leading to signs of congestive cardiac failure an uncommon presentation for neonates with TOF physiology Management of such cases requires both immediate and long-term considerations. Stabilization with oxygen therapy, PDA maintenance, and diuretics may address acute symptoms, while surgical intervention or catheter-based therapies will likely be necessary for definitive management. [11,12] Early recognition of this rare combination of defects is critical to avoid delays in treatment, which could lead to significant morbidity or mortality. This case highlights the importance of comprehensive echocardiographic evaluation in neonates presenting with cyanosis and respiratory distress. The findings of PA-IVS with TOF-like physiology and PR emphasize the need for individualized management strategies. The long-term outcomes and pathophysiological mechanisms underlying this unique combination of congenital anomalies needs to be explored studied in depth. [13]

4. CONCLUSION

Any baby with TOF physiology with unusual TOF and signs of congestive cardiac failure the component of pulmonary regurgitation should be kept in mind and managed accordingly. Patients with TOF physiology typically do not present with congestive cardiac failure. The co-occurrence of PR in a neonate with PA-IVS and TOF-like physiology is an unusual finding that requires further investigation. This case underscores the complex interplay of anatomical and physiological factors contributing to the rare presentation and highlights the need for research into long-term outcomes and mechanisms of PR in such settings.

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