

CASE REPORT

Tetralogy of Fallot in a child with VACTERL association - A case report of uncommon occurrence

Authors

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ABSTRACT:

Tetralogy of Fallot (TOF) is the most common form of cyanotic congenital heart disease. The VACTERL acronym stands for the non-random association of vertebral, anal, cardiac, tracheoesophageal, renal, and limb anomalies in a single patient. Approximately two-thirds of this rare sporadic disorder have cardiac anomalies, the common being the ventricular septal defect and atrial septal defect. We report a case of rare combination i.e, VACTERL association with TOF.

KEYWORDS: Tetralogy of Fallot, VACTERL association, ventricular septal defect

1. CASE:

A 12 yr old boy was brought with complaints of shortness of breath and palpitations on exertion. There was a history of squatting episodes pointing towards decreased pulmonary blood flow conditions. The prenatal and perinatal history of the mother during the pregnancy was normal. He attained developmental milestones at a normal age as his peers. He was underweight for his age with a mid-arm circumference of 12.0 cm. The physical examination revealed dysmorphic features i.e., low set ears, hypoplastic mandible, deciduous teeth, left

absent thumb, radial hypoplasia (figure 1), kyphoscoliosis (figure 2) and presence of pandigital clubbing. He was afebrile with a saturation of 80% on room air. The heart rate was 84 per min, and his blood pressure was normal for age. The jugular venous pressure was not elevated. Cardiovascular examination revealed parasternal heave of grade 3, systolic thrill in left second and third intercostal space. The second heart sound was single with an absent pulmonary component. There was a grade 4/6 mid systolic murmur at the left 2nd and 3rd intercostal space. The other systemic examination was normal.

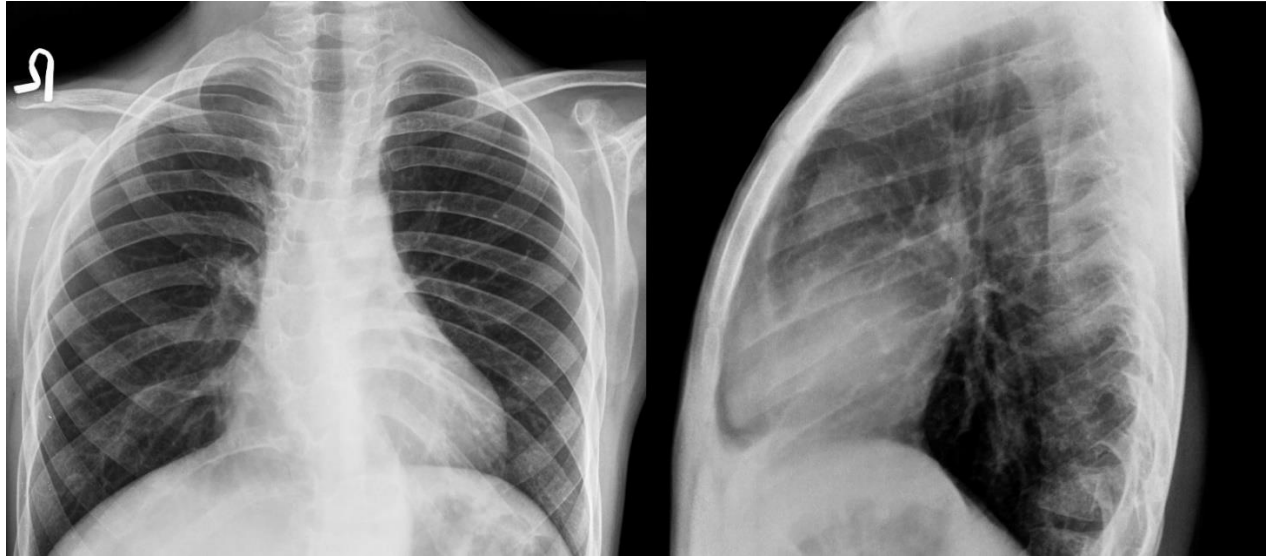


Figure 1: (Left image) Chest X-ray PA view showing normal cardiac silhouette with pulmonary oligemia. There is scoliosis of the thoracic spine. (Right image) Lateral view showing right ventricular hypertrophy with the obliteration of the retrosternal space, with the narrowed pulmonary artery. Retrocardiacally, there is oligemia of the pulmonary fields.

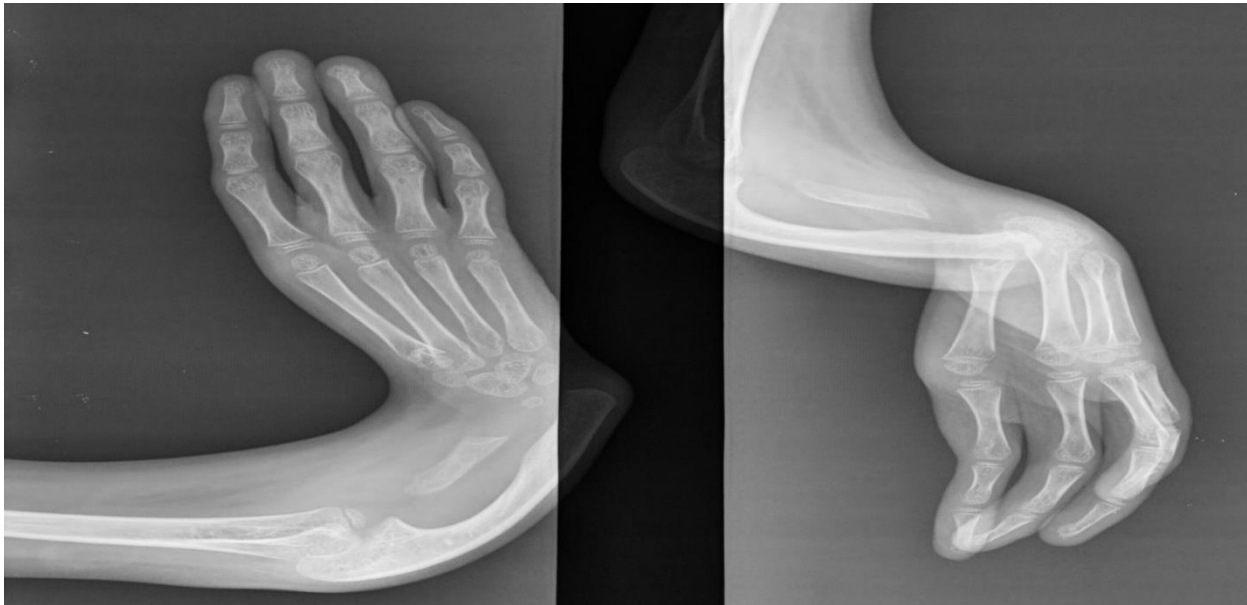


Figure 2: X-ray of the forearm anteroposterior (left) and lateral view (right) showing radial hypoplasia and absent thumb.

His biochemical profile showed hemoglobin of 13gm/dl, normal total leukocyte count, and normal electrolytes. The electrocardiogram showed sinus rhythm, right axis deviation, and right ventricular hypertrophy. The chest x-ray showed upturned cardiac apex, oligemic lung fields, kyphoscoliosis of the thoracic spine (figure 2a). The right ventricle was enlarged with the obliteration of lower retrosternal space on the lateral view (figure 2b). The pulmonary trunk and infundibular region were narrowed (figure 2b). The echocardiogram showed situs solitus. There was a large perimembranous ventricular septal defect of 1.3 cm (Figure 3). (bidirectional shunt). There was 50% overriding of the aorta over the defect (Figure 3, lower right), and infundibular hypertrophy as a result of the malaligned septum. The

pulmonary valve was doming and the main pulmonary artery was hypoplastic. The peak pressure gradient across the pulmonary valve (Pmax) was 92mm Hg with a peak velocity (Vmax) of 4.8 m/sec (Figure 4, Upper Right). There was a significant dynamic obstruction at the infundibular level represented by the characteristic dagger-shaped pattern on the spectral Doppler (Vmax 3.4m/sec, Pmax/Pmean 47/14 mm Hg) (Figure 4, lower left).. The right ventricle was hypertrophied with normal function. The left ventricular function was normal(Figure 4, lower right). There were no other defects. The aortic arch was left-sided and the arch vessels were normal. The ultrasound abdomen showed a horseshoe-shaped kidney with normal renal function tests.

He was kept on propranolol 10 mg bid. He improved symptomatically and had a

significant decrease in gradients when assessed after three months of follow up.

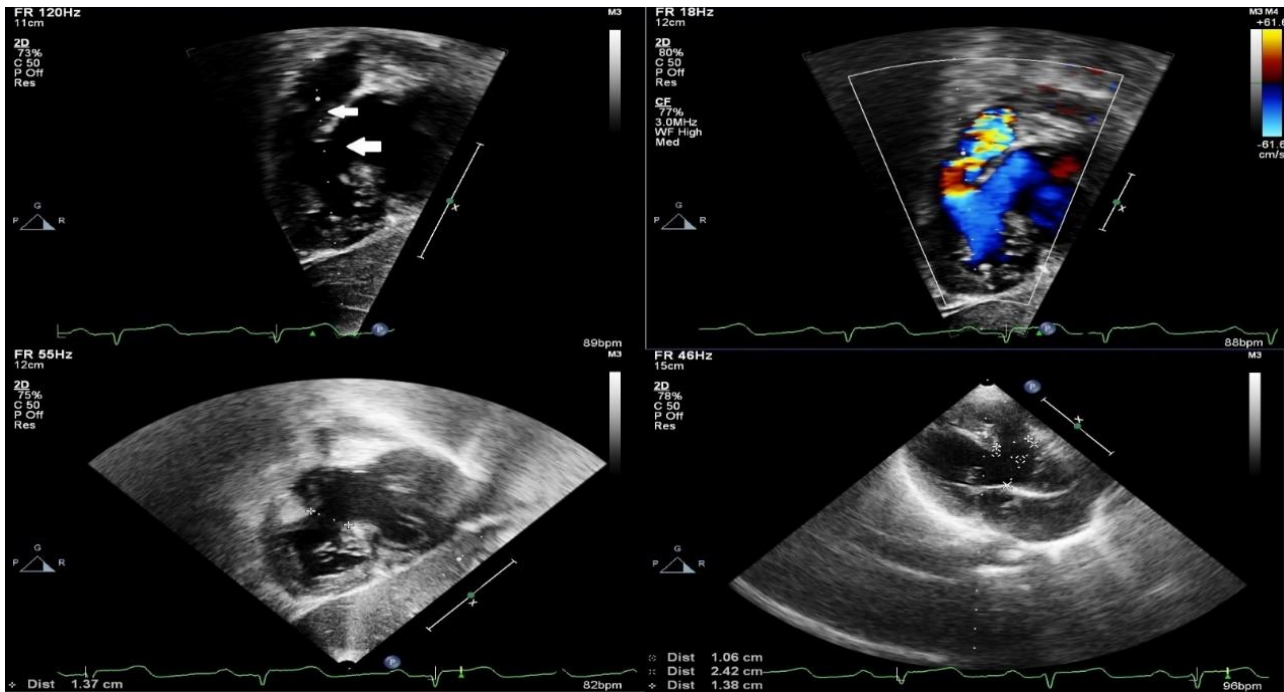


Figure 3: Echocardiographic images; (upper left) Subcostal view showing the ventricular septal defect and turbulence in the pulmonary artery (upper right). The lower left image showing a large ventricular septal defect of 1.37cm and lower right image showing the 50% overriding of the aorta over the ventricular septal defect.

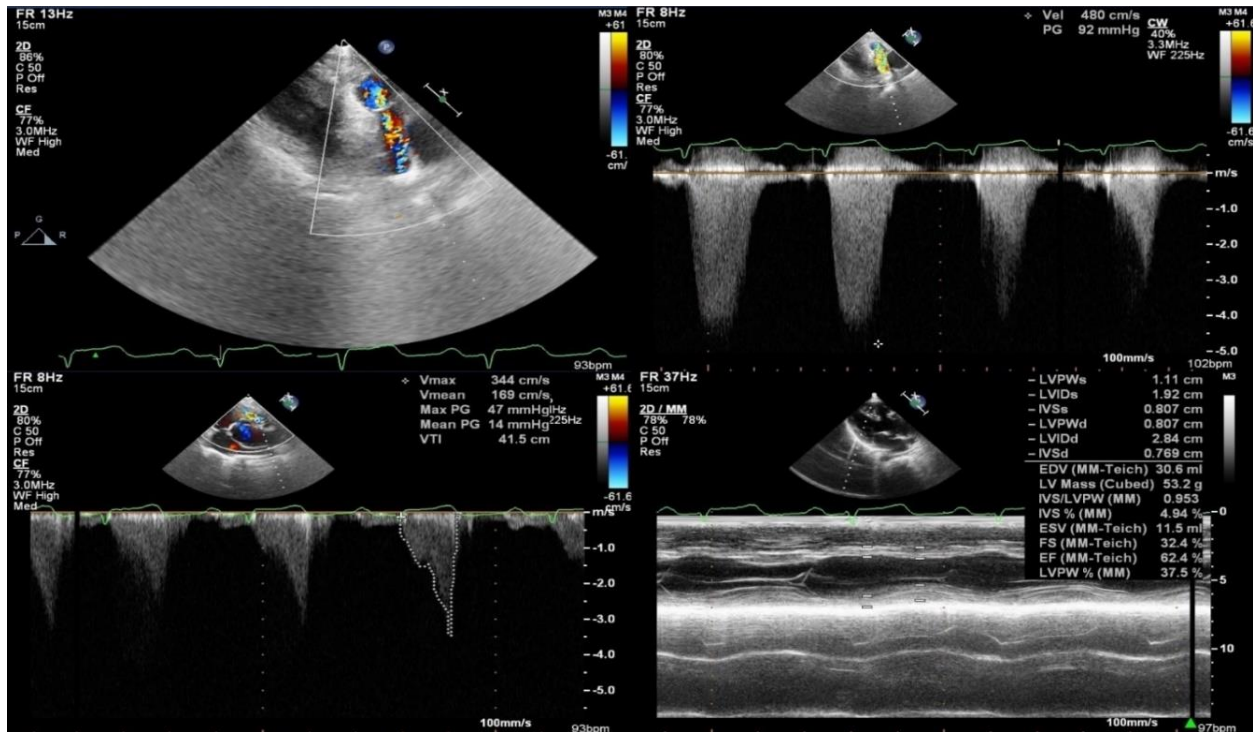


Figure 4: Echocardiographic images; (upper left) Parasternal short axis view showing the turbulence at the level of the pulmonary artery. (upper right) continuous-wave spectral Doppler image with a gradient of 92 mmHg across the pulmonary valve. (lower left) pulse wave Doppler image showing dagger-shaped configuration at the level of infundibulum with a peak gradient of 47 mm Hg. (lower right) Normal left ventricular function on M mode.

2. DISCUSSION:

Association refers to the non-random co-occurrence of a group of multiple malformations rather by chance and not implying a single underlying cause or being components of known syndrome.¹ The classical association disorder is the VATER association, first described by Quan and Smith in 1973.² The occurrence of the disorder is rare with an incidence of 1 in 10,000 to 40,000 newborns.¹ The acronym

VATER stands for V – vertebral anomalies, A – Anal atresia, TE – Tracheoesophageal fistula with esophageal atresia, R – radial, and renal anomalies. The term VACTERL to include C for cardiac anomalies and L for limb defects was proposed by Nora et al.³

There are no validated diagnostic criteria to date for the diagnosis and no laboratory test to either diagnose or rule out the VACTERL association. Traditionally for a diagnosis of VACTERL association to be made there

should be three features of the six abnormalities.⁴ As per the researchers, a “complete” diagnosis of VACTERL association is made if there is the presence of at least one anomaly in each of the three body parts (i.e., limbs, thorax, and pelvis/lower abdomen), and “probable” in presence of two or more anomalies in two body parts. The diagnosis requires a complete physical examination as it is based on the defects present in each body part. The tests can be performed based on the suspected birth defects.⁵

In the VACTERL association, the vertebral (hemivertebrae or hypoplastic vertebrae) and the trachea-oesophageal fistula with esophageal atresia are the common anomalies. They occur in 70-80% of cases. The limb anomalies (absent thumb, polydactyly, syndactyly, and forearm defects) vary between 50-70%. The next common after the above in occurrence is the presence of imperforate anus and renal anomalies (renal atresia, horseshoe kidney) seen in approximately 50%. The cardiac anomalies vary between 40-60%. The common cardiac anomaly is a ventricular septal defect (50-70%) and then is the atrial septal defect (30%).⁶ The occurrence of

Tetralogy of Fallot (TOF) is 16% as per Weaver et al.⁷

Etiology remains unknown, although familial clustering is seen. There is a hypothesis of association with genes of the SHH (Sonic Hedgehog) pathway during blastogenesis causing the deformities.⁸ Because of the blastogenic origin and abnormalities occurring at various levels, Martinez- Frias et al termed polytopic field defects rather than them as a simple association.⁹

Although the children with VACTERL association can have complications due to the malformations or secondary to surgery, they are not life-threatening (horseshoe kidney, radial hypoplasia).

The treatment is directed towards the symptomatic specific malformations (tracheoesophageal fistula, imperforate anus) in an individual. The presence of a tracheoesophageal fistula, imperforate anus requires surgery in the neonatal period. There can be a requirement of repeat surgeries in the latter part of life. A team approach (physician, surgeon, gastroenterologist, anesthetist, cardiologist, physiotherapist, vascular surgeon, neurosurgeon, orthopaedician, etc.) for effective management of individual cases with

VACTERL association. They need close follow up..

Our patient had four features of the VACTERL association i.e., vertebral (kyphoscoliosis of the thoracic spine), cardiac (Tetralogy of Fallot), renal (horseshoe kidney), and the limb anomalies (radial hypoplasia, absent thumb). The closest differential diagnosis is Holt Oram syndrome. The facial, auditory, ocular, vertebral and renal involvement rules out the possibility of the Holt-Oram syndrome.¹⁰

3. CONCLUSION:

The cardiac anomalies in the VACTERL association account for 40-60% of cases. The ventricular and atrial septal defects are common cardiac anomalies. The occurrence of TOF is relatively rare and is hence being reported.

4. ACKNOWLEDGMENTS:

None

5. CONFLICTS OF INTEREST:

None declared by the authors

Contribution by the authors:

All the authors are affiliated to the Department of Cardiology, Osmania General Hospital, Hyderabad though present affiliations have been changed for AKK, MVK, SRCh. PN, AKK admitted and evaluated the case. PN, AKK have been assisted by MVK, SRCh during the echocardiography and the literature review of the cardiac defects in the VACTERL association. The manuscript was supervised by the RS, OAN, KMKP. All authors have agreed on PN as the corresponding author, who has edited the manuscript. The final manuscript was read and approved by all the authors before submission to the journal. The final proof check was read by all the authors.

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