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Case Report

Adult Tetrology of Fallot: A Doppler Echocardiographic Finding

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Abstract: In TOF the morphological abnormalities are – overriding of aorta, pulmonary obstruction, right ventricular hypertrophy and ventricular septal defect. Normally it is nonrestrictive means free communications between the ventricles but sometimes very rarely it can become restrictive when the tricuspid valve restrict the flow across the ventricular septal defect. Among children with CHD 10% of them report TOF. Chest radiographs usually show a normal-size heart silhouette, with an upturned apex and a concave main pulmonary artery segment, commonly known as "boot-shaped" heart. On the electrocardiogram, it is common to see signs of right atrial enlargement and right ventricular hypertrophy showing right axis deviation, prominent R waves anteriorly and S waves posteriorly, upright T wave in V1 (abnormal after 7 days of life up to 10 years of age) and a qR pattern in the right precordial leads. If the ration between pulmonary artery orifice diameter to aortic orifice diameter is <.3 primary repair is unsuccessful and in that case we must go for shunt surgeries which are palliative procedures till permanent repair can be done. This should add proper assessment of coronary artery origin. This is a case of adult tetralogy of fallot (TOF) coming to outpatient department of cardiology with complaints of chest discomfort and sometimes cyanotic spells. Age of the patient is 42 years male. Doppler Echocardiography was done. In the image overriding of aorta was found around 20% over interventricular septum. Left sided aortic arch was detected along with ventricular septal defect (VSD) with size 14 mm. Size of pulmonary orifice 10 mm and that of aortic orifice was 22 mm. Hence pulmonary artery orifice was found to get stenosed.

Keywords: Tetralogy, Aorta, Artery, Orifice, Ventricle, Chest

1. Introduction

In TOF the morphological abnormalities are – overriding of aorta, pulmonary obstruction, right ventricular hypertrophy and ventricular septal defect. There are two types of TOF depending upon the flow across the VSD, non restrictive where there is free flow across the VSD and restrictive where the tricuspid valve restrict the flow across the VSD. There are usually four types of TOF.

1) TOF with pulmonary atresia
2) TOF with absent pulmonary valves
3) TOF with double outlet right ventricle
4) TOF with atrioventricular septal defect. [1] The incidence of TOF is 1 in 3500 live births and incidence is equal in both male and female. Cases are usually sporadic. There is about 3% risk of familial recurrence. [2] Cytogenetic abnormality detected so far in TOF is trisomies 21, 18, 13. And microdeletion of chromosome 22. TOF with pulmonary atresia is mostly associated with mcrodeletion. Other deformities associated with microdeletion are palatal deformities, deformed facies, immunodeficiency, hypocalcaemia, learning disability which is frequently termed as DiGeorge syndrome. [9] Other chromosomal abnormalities detected were 47, XXX & 47 XYY, microduplication of chromosome Y, 47 XX and 47 XY, deletion of chromosome 5, 6 & t (10;13) & t (1;9). These trisomies and translocations may be found isolated or in combinations. These chromosomal aberrations and chromosomal rearrangements were confirmed by Chromosomal microarray analysis. [10, 3]In 4% of tetralogy of Fallot patients there is mutation of NKX2.5 gene.; in Alagille syndrome there is higher

incidence of tetralogy of Fallot which involves mutation of JAG1 gene; a few patients in holt Oram syndrome have mutation of TBX5 with tetralogy of Fallot and FOXC2 in hereditary lymphedemadistichiasis, in which rare patients also have tetralogy of Fallot. In chromosome 22 there may be found microdeletion of TBX5 gene which may account for 15% cases of TOF. Trisomy 21, 18, and 13 account for 10% of tetralogy of Fallot cases and genes responsible for it are still to identify. [4] Among these Allagilli syndrome and DiGeorge syndrome are autosomal syndromic TOF. There are autosomal nonsyndromic TOF genes likeNKX2.5, GATA4, GATA5, GATA6, ZFPM2, GDF1, TBX1, GJA5 [7] Steno of Denmark first described the defect in 1673, and Fallot of Marseilles coined the term tetralogy in 1888. [5] Co-associated cardiac anomalies are right aortic arch, present or absent of ductus arteriosus, persistent left superior venacava, endocardial cushion defect, aberrant right subclavian artery, left pulmonary artery sling, major aortopulmonary collaterals, tricuspid atresia. Among these right aortic arch is most common followed by present or absent ductus arteriosus. The noncardiac anomalies are-hypoplasia of the nasal bone, short long bone, fetal growth restriction, single umbilical artery, hypoplasia of the thymus, nuchal fold thickening (≥6mm), ventriculomegaly, heterogeneous echo pattern of liver, hypoplasia of phalanx, echogenic bowl, echogenic kidneys, club foot, omphalocel, low-set ears, overlapping fingers, arachnoid cyst, micrognathia, ectrodactylia, cystic hygroma, strawberry-shaped skull, long-eyebrows, cerebellar hypoplasia, hypoplasia of the lung, esophago-tracheal fistula, exencephaly, hemivertebra. [6] Majority of patients with TOF die due to severe right ventricular outflow tract obstruction. Severity of obstruction increases with advancement of age. If uncorrected 25% die with cyanotic spells by 1st year of life, 40% by 3years of age, 70% by 10years of age, 95% by 40 years of age. 50% of patients are diagnosed antenatally by fetal echocardiography, requiring for postnatal prostaglandin therapy if there is evidence of severe right ventricular outflow obstruction. Other useful tools are chest radiograph, electrocardiogram, and echocardiogram. Chest radiographs usually show a normal-size heart silhouette, with an upturned apex and a concave main pulmonary artery segment, commonly known as “boot-shaped” heart. On the electrocardiogram, it is common to see signs of right atrial enlargement and right ventricular hypertrophy showing right axis deviation, prominent R waves anteriorly and S waves posteriorly, upright T wave in V1 (abnormal after 7 days of life up to 10 years of age) and a qR pattern in the right precordial leads, [8] If the ration between pulmonary artery orifice diameter to aortic orifice diameter is <.3 primary repair is unsuccessful and in that case we must go for shunt surgeries which are palliative procedures till permanent repair can be done. This should add proper assessment of coronary artery origin. The different shunt surgeries are- Pott shunt, blalock-taussing shunt, Waterson shunt. [11] With advancement of age from<1yera to>5 years in patients with TOF there is increase in incidence of myocardial fibrosis resulting in right ventricular myocardial dysfunction, 16]In pentalogy of fallot echocardiography shows large sub-aortic perimembranous ventricular septal defect (VSD) with 50% aortic override, large ostium-secundum atrial septal defect (ASD), thickened septal leaflet of the tricuspid valve with severe tricuspid regurgitations, pulmonary valve thickening, dilated and hypertrophied right ventricle, non visualization of coronary arteries, 17].

2. Case Report

This is a case of adult tetralogy of fallot (TOF) coming to outpatient department of cardiology with complaints of chest discomfort and sometimes cyanotic spells. Age of the patient is 42 years male. Doppler Echocardiography was done. In the image overriding of aorta was found around 20% over interventricular septum. Left sided aortic arch was detected along with ventricular septal defect (VSD) with size 14 mm. Size of pulmonary orifice 10 mm and that of aortic orifice was 22 mm. Hence pulmonary artery orifice was found to get stenosed.

3. Discussion

Singh B. and Mohan. J. C. in 1992 found normal aortic orifice to be 15-23 mm in size and pulmonary orifice 15-22 mm in normal Doppler echocardiography. Here in this case pulmonary orifice diameter is 10 mm so stenosed. [12]

According to the finding of Awashthy N, and S. Radhakrishnan in 2013 ventricular size can be determined from Doppler echocardiography. VSD size is mild if it is 1/3rd of aortic size, moderate if 1/3rd -2/3rd of aortic size and large if equal to aortic orifice diameter. VSD (14 mm) in this case is found to between 1/3rd -2/3rd to that of aortic orifice diameter (22 mm), so moderate VSD. [13]

Maria S. C. and Branidou K. in 2008 mentioned a case with TOF diagnosed in around 75 years of age when he came for checkup complaining of dyspnea and cyanotic spells. There was 50% overriding of aorta on ventricular septum, 22mm size VSD and right ventricular infundibular obstruction (pulmonary stenosis). Here the aorta is overriding 20% of the interventricular septum and the patient is now complaining of cyanotic spells with dyspnea as he was apparently normal before. [14]

Craatz S., Kunzul E., Spanel Borowski K. in 2003 mentioned that in TOF due to overriding of aorta when there is pulmonary stenosis the aortic arch is right sided. In our case the arch is left sided as it is overriding only 20% of interventricular septum. [15]

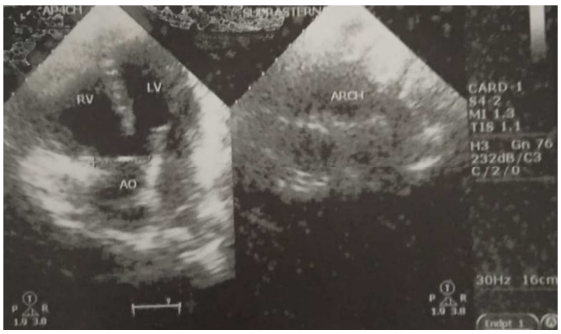

Figure 1. *Doppler echocardiograpgy showing TOF with RV-right ventricle, LV-left ventricle, AO- aortic opening, ARCH- aortic arch.*

4. Conclusion

Doppler echocardiography may be a diagnostic tool for a case of sudden onset cyanotic spell with dyspnea with the help of which cardiac surgeons can plan for palliative surgical procedures till a parmanent remedy

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