Tetralogy of Fallot with Quadricuspid Aortic Valve

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Abstract
We describe herein a 54 year female who had tetralogy of Fallot with quadricuspid aortic valve. This combination is very uncommon. Hence it was worth reporting this interesting case.

Introduction
Tetralogy of Fallot (TOf) is the most common cyanotic congenital heart disease, usually accompanied by right sided aortic arch, patent ducus arteriosus, pulmonary atresia, aortic regurgitation, persistent left superior venacava and anomalies of the coronary arteries. The association with quadricuspid aortic valve (QAV) is seldom reported. We present here a case of TOf with QAV.

Case Report
A 54 year female presented in emergency department with complains of breathlessness on exertion and cyanosis. On examination her pulse was 88/mt regular, blood pressure - 100/56 mmHg and respiratory rate - 22/mt Central and peripheral cyanosis with bilateral clubbing (grade III) was evident on fingers and toes. However 'a' wave in jugular venous pulse was not prominent. Apex beat was subxiphoid in location and retractile formed by right ventricle. There was a palpable thrill in left third and fourth intercostal space parasternally. On auscultation S 1 was normal whereas S 2 was single. An ejection click was audible in right second intercostal space. There was an ejection systolic murmur in pulmonary area and a diastolic murmur of grade 4/6 at Erb’s area. Hematologic findings with blood biochemistry, renal and liver function tests were within normal limits. On measurement of arterial partial oxygen pressure oxygen saturation was 88%. Chest skiagram revealed normal sized, ‘boot shaped’ heart with pulmonary oligaemia. Electrocardiogram revealed right axis deviation with peaked ‘P’ waves and sudden transition of prominent ‘R’ wave in lead V 1 to dominant ‘S’ wave in lead V 2 (Figure 1). Finally transthoracic echocardiography was done which revealed non-restrictive ventricular septal defect (VSD) of 4cm size, overriding aorta to the right and anteriorly (>50%), right ventricular hypertrophy (12mm) with infundibular right ventricular outflow obstruction (gradient-66mmHg) (Figure 2). An additional characteristic echocardiographic feature was the presence of a quadricuspid aortic valve in short axis parasternal view having a cross shape in diastole with one large, one small and two intermediate cusps and severe aortic regurgitation jet directed into the right ventricle (RV) (Figure 3). Patient was advised surgery but he refused and was treated medically with diuretics, digoxin and was discharged after 1 week with symptomatic improvement.

Discussion
QAV is a rare congenital anomaly with overall incidence of 0.01%. It is often associated with other cardiac disorders such as patent ductus arteriosus, VSD, pulmonary and subaortic stenosis, coronary anomalieshypertrophic cardiomyopathy and congenital complete heart block. To the best of our knowledge there are, so far only three reported cases of QAV with TOf. Hurwitz and Robert classified QAV into seven different types and named them A to G. Type ‘A’ having all four equal aortic cusps is the commonest and type ‘E’ with three equal and one large cusps is the rarest. Our patient has a type ‘D’ QAV, represented by one large, one small and two intermediate cusps; leading to severe aortic regurgitation (AR) owing to their unequal size. The findings reported in the literature suggest that fibrous thickening of valves, along with unequal distribution of mechanical stress

Fig. 1 : Electrocardiogram showing RAD, p-pulmonale and sudden transition of prominent ‘R’ wave in lead V 1 to dominant ‘S’ wave in lead V 2

Fig. 2 : Transthoracic echocardiogram (PLAX view) showing large non-restrictive VSD with over-riding aorta
on the valve, may lead to progressive AR. In our patient the incompetent aortic valve constituted hemodynamic burden for the RV, as the regurgitant jet was directed into the RV through the nonrestrictive VSD, contributing to it’s dilation due to longstanding volume overload. There is a strong possibility that the presence of chronic AR was somehow beneficial, by raising the proportion of oxygenated blood in the RV. As a consequence, the right to left shunt provided the left ventricle with partly oxygenated blood, resulting in a lower than expected degree of cyanosis and hypoxic damage to the myocardium, which enabled prolonged survival in our patient.

References