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Double Outlet Right Ventricle With Intact Ventricular Septum And Interrupted Aortic Arch: A Case Study Of A Foetus With Trisomy 18

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Abstract:
Background: *90% of the infants with trisomy 18 suffers from congenital heart defects. Double outlet right ventricle (DORV) is a rare form of congenital heart disease where both the arterial roots arise from the right ventricle. Interrupted aortic arch (IAA) is an extremely rare congenital anomaly representing approximately 1% of congenital heart disease, characterised by lack of luminal continuity between the ascending and descending thoracic aorta*
Case Report: *A still born male foetus of around 20 weeks gestation, weighing 238g with trisomy 18 was collected from the Obstetrics and Gynaecology Department, RIMS, Imphal. On dissection, both the great arteries are committed to the right ventricle and are unobstructed. The aorta arose right to the pulmonary trunk and is interrupted distal to the left subclavian artery. Pulmonary-aortic connection is maintained by a large patent ductus arteriosus. The mitral valve was atretic. The left ventricle was hypoplastic.*
Conclusion: *We found this case notable as this is an exceedingly rare case of DORV with intact ventricular septum, in the setting of mitral atresia, hypoplastic left ventricle associated to type A, IAA in Trisomy 18*.
Key Words: *Double outlet right ventricle, ventricular septum, ventricular septal defect, interrupted aortic arch, mitral atresia, hypoplastic left heart, trisomy 18.*

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I. Introduction

Trisomy 18 (47, XY + 18), also known as Edwards syndrome, is the second most common autosomal trisomy after trisomy 21 and is caused by meiotic non-disjunction, an error in cell division producing different phenotypic outcomes throughout the body. The incidence of trisomy 18 is estimated to be about 1 per 6000 live births.[1]

Infants with trisomy 18 have multisystem congenital malformations, out of these congenital heart defects occur in about 90% of the children.[1] Double outlet right ventricle (DORV) is a rare form of congenital heart disease where both the arterial roots arise from the right ventricle. Ventricular septal defect (VSD) is present in almost all the cases and represents the only outlet from the left ventricle. DORV with intact ventricular septum is rare.[2,3] Interrupted aortic arch (IAA) is an extremely rare congenital anomaly representing approximately 1% of congenital heart disease which is characterised by lack of luminal continuity between the ascending and descending thoracic aorta.[4] There are three types of IAA: Type A is interrupted aortic arch distal to the left subclavian artery. Type B, between the left common carotid and the left subclavian arteries. Type C, between the innominate and left common carotid arteries.[5,6] IAA is often found with other defects such as patent ductus arteriosus, ventricular septal defect, transposition of great arteries and aortic stenosis.[6] Occurrence of hypoplastic left ventricle along with DORV and mitral atresia are part of the syndrome of "mitral atresia with patent aortic root" as clarified by Mickell et al. in 1983.[7] Described here is an example of DORV with intact ventricular septum, hypoplastic left ventricle with mitral atresia and interrupted aortic arch in trisomy 18.

II. Case Report

A still born male foetus of around 20 weeks of gestation with trisomy 18 (Edward's syndrome) with the maternal age of 40 years of fourth gravida was collected from the Obstetrics and Gynaecology Department, RIMS, Imphal, after taking consent from the parents and permission from the Research Ethics Board, RIMS,

Imphal bearing reference number: A/REB/Props(SP)208/184/25/2023. The study was carried out in the Department of Anatomy, RIMS. Antenatal quadruple test reveals increased risk for trisomy 21 (1:168), trisomy 18 (1:5). Edward's syndrome was detected on Amniocentesis and QFPCR (Quantitative Fluorescent Polymerase Chain reaction). On antenatal ultrasonographic soft markers there was presence of bilateral choroid plexus cyst, bilateral club foot and hypoplastic left heart.

The foetus was then subjected to external examination, radiological examination and gross anatomical dissection. The crown rump length (CRL) of the foetus was 14.9cm weighing 238g. The cephalic index of the said foetus was 88.03. On external examination, there was presence of bilateral clenched hand with overlapping of the index finger on the right side and bilateral rocker-bottom feet. The skeletal system appears to be normal on CT (computed tomography) 3D reconstruction.

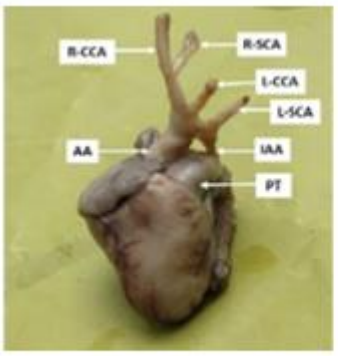

FIG.1: Anterior veiw

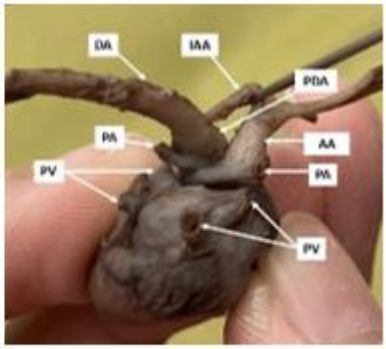

FIG.2: Posterior veiw

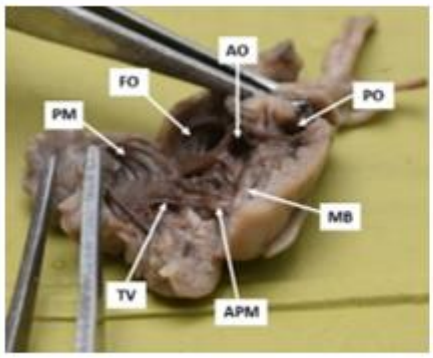

FIG.3: Interior of right ventricle

FIG.4: Hypoplastic left ventricle

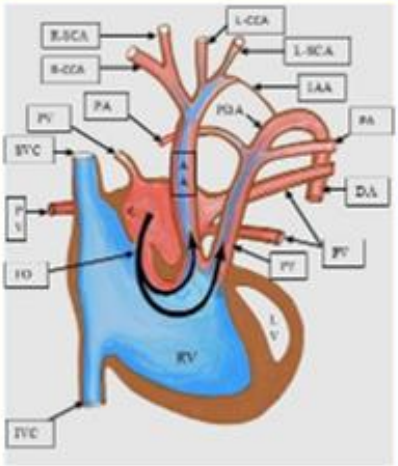

FIG.5

FIG.1&2: Exterior of the heart. AA: Ascending aorta, DA: Descending aorta PT: Pulmonary trunk, R-CCA: Right common carotid artery, L-CCA: Left common carotid artery, R-SCA: Right subclavian artery, L-SCA: Left subclavian artery, IAA: Interrupted aortic arch, PA: Pulmonary artery, PV: Pulmonary vein, PDA: Patent ductus arteriosus.
FIG. 3&4: Interior of the heart. PM: Pectinate muscle, FO: Foramen ovale, AO: Aortic opening, PO: Pulmonary opening, MB: Moderator band, APM: Anterior papillary muscle, TV: Tricuspid Valve, LV: Left ventricle
FIG.5: Schematic diagram of the heart. AA: Ascending aorta, DA: Descending aorta PT: Pulmonary trunk, R-CCA: Right common carotid artery, L-CCA: Left common carotid artery, R-SCA: Right subclavian artery, L-SCA: Left subclavian artery, IAA: Interrupted aortic arch, PA: Pulmonary artery, PV: Pulmonary vein, PDA: Patent ductus arteriosus, IVC: Inferior vena cava, SVC: Superior vena cava, FO: Foramen ovale, LV: Left ventricle, RV: Right ventricle

The foetus was then dissected and examined for any internal deformity. Usual arrangement of the abdominal and the thoracic organs were found with the heart in the left hemithorax. Both the great arteries were committed to the right ventricle and were unobstructed. The aorta arose right to the pulmonary trunk and was interrupted right after giving out the left subclavian artery (Fig:1 and Fig:2). Pulmonary trunk appears dilated and the continuity of aorta was maintained by a large patent ductus arteriosus (PDA), which is a pulmonary-aortic connection. The mitral valve was atretic and the only egress of the left atrium was through foramen ovale. The dominant anterior ventricle, where both the aorta and pulmonary artery open, appear to be of right ventricular morphology due to the presence of a moderator band (Fig:3) and the ventricular septum was found to be intact. The left ventricle was hypoplastic and slit like (Fig:5).

III. Discussion

The foetus was small for its age as we compare the CRL and weight with that of the normal foetus which is 19 cm and 460 g respectively for a foetus with same gestational age.[8] All the external malformations described, as well as that of those found in USG soft markers were characteristic findings of trisomy 18.[9]

Congenital heart disease occurs in about 90% of patients with trisomy 18, the most common being VSD.[1] Double-outlet right ventricle with intact ventricular septum is a rare occurrence which reflects closure of a previously existing defect, particularly when associated with absence or hypoplasia of the left ventricle.[10] The left ventricle played no role in maintaining the systemic arterial circulation since the left atrioventricular connection was absent and the only egress to the left atrium was through foramen ovale. The only connection to the descending aorta was maintained by the patent ductus arteriosus, as there was a presence of a fibrous band with no lumen between the aortic arch distal to the left subclavian artery and distal part of the PDA. Since the interruption of the aortic arch was right after the left subclavian artery it is type A, IAA.[5] Direct fibrous continuity between aortic and tricuspid valve is due to normal absorption of sub aortic conus. Mitral atresia and left ventricular underdevelopment may lead to double outlet right ventricle relation, rather than resulting from malformation of the infundibulum and great arteries.[3]

DORV with intact ventricular septum and interrupted aortic arch is an extremely rare combination of cardiac malformation which has been described in limited number of literatures so far as described by Karmegaraj et al.[11] Few reports of DORV with mitral atresia, however, have been described in association with trisomy 18.[3,10]

IV. Conclusion

We found this case notable for its rare combination of complex cardiac malformations and the presence of interrupted aortic arch. This is an exceedingly rare case of DORV with intact ventricular septum, in the setting of mitral atresia, hypoplastic left ventricle associated to type A, IAA in Trisomy 18. Early diagnosis and prompt treatment of IAA and its related cardiovascular anomalies is a multidisciplinary approach which is required for better prognoisis.[10] So, the present study requires understanding of complex events of development of heart and aortic arches in trisomy 18.

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