A Rare case report of heterotaxy syndrome
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Abstract: Heterotaxy syndrome is the Pattern of malformation involving multiple organs and occurs as a result of failure of lateralization of thoracic and abdominal viscera into either a normal pattern (situs solitus) or inverted pattern (situs inversus). We report a rare case with prenatal diagnosis of complex cardiac abnormalities associated with left isomerism. This is a case of term female baby admitted with respiratory distress and cyanosis since birth. Post natal investigations confirmed heterotaxy syndrome with asplenia. Baby treated with symptomatic measures and referred to further higher centre for intracardiac repair.

Keyword: Heterotaxy, isomerism, fetal echocardiography, intracardiac repair

Case report:
B/O Saranya, first order female child born to non consanguineous parents delivered by caesarean section at government Rajaji hospital Madurai with prenatal sonography showing evidence of situs laterality. Baby was admitted in neonatal intensive care unit with respiratory distress and cyanosis since birth. On examination, baby had cleft lip, apical beat in right hemithorax, with oxygen saturation of 82%. The following investigations were done: chest radiograph revealed dextrocardia, cardiomegaly with liver on left side and stomach bubble on right. Ultrasound abdomen showed liver with normal echoes and absence of spleen. Echocardiogram revealed dextrocardia, common atrium, common atrioventricular canal, large membranous annular ventricular septal defect, aorta arising from left ventricle, pulmonary atresia and major aorto pulmonary collaterals. ECG showed normal axis. Peripheral smear showed no evidence of Howell Jolly bodies or RBC inclusions. Baby was treated with antibiotics and other supportive measures. Cardiothoracic opinion obtained suggested intracardiac repair. Hence baby was referred on the 10th day of life to higher centre for further management.

Ivemark Syndrome is a multiple organ syndrome associated with splenic abnormalities, complex cardiac pathology and an abnormality of the abdominal viscera. Etiology inheritance is Autosomal recessive (most cases), autosomal dominant, or X-linked inheritance. Heterotaxy syndromes could also occur in chromosomal translocation or deletion in sporadic cases. Etiology remains undetermined. Layton's hypothesis suggests lack of normal genetic control leading to random organization of viscera. Mutation in connexin 43 gap junction gene leads to abnormal cell cell communication causing Viscero atrial heterotaxy. Embryology: During 5th week of gestation major events in normal cardiac development occurs-the Cardiac tube undergoes septation, there is growth of endocardial cushions, separation of conotruncus and lobation of lungs occur. Rotation of gut occurs between 28th-35th day. Connection between left atrium and pulmonary venous plexus occurs between 30th-32th day. Spleen arises from left mesogastrium between 35th-36th day. Terminology The term situs refers to the position of the atria and viscera relative to the midline. The atrium whose appendage is broad-based and receives blood from the inferior vena cava may be called the systemic or right atrium. The atrium with the smaller, narrower appendage and that receives blood from the pulmonary veins is called the pulmonary or left atrium. Situs solitus is the usual arrangement of organs and vessels within the body. The systemic atrium is on the right with a right-sided trilobed lung, liver, gallbladder, and inferior vena cava. The pulmonary atrium is on the left with a left-sided bilobed lung, stomach, single spleen, and aorta. The cardiac apex is on the left.
The incidence of congenital heart disease in patients with situs inversus and levocardia is only 0.6%–0.8%. Situs inversus refers to an anatomic arrangement that is the mirror image of situs solitus. The systemic atrium is on the left with a left-sided bilobed lung, liver, gallbladder, and inferior vena cava. The pulmonary atrium is on the right with a right-sided bilobed lung, stomach, single spleen, and aorta. The cardiac apex is on the right. Situs inversus is seen in 0.01% of the population, and the incidence of congenital heart disease in patients with situs inversus is 3%–5%. Situs ambiguous, or heterotaxy, refers to visceral malposition and dysmorphism associated with indeterminate atrial arrangement-ment. This abnormal arrangement of body organs is different from the orderly arrangement seen in situs solitus or situs-versus. The incidence of congenital heart disease in patients with heterotaxy is very high, ranging from 50% to nearly 100%. Heterotaxy Syndrome with Asplenia. This syndrome occurs more frequently in males, and patients often present with cyanosis and severe respiratory distress. The designation of classic right isomerism or bilateral right-sidedness implies that the patient has bilateral trilobed lungs with bilateral minor fissures and epaerterial bronchi, bilateral systemic aorta, a centrally located liver, and a stomach in indeterminate position. Cardiac malformations include Dextrocardia (30-40%), pulmonary blood flow, TGA (60-75%), TAPVC(70-80%), Common AV valve (80-90%), Single ventricle (40-50%), Bilateral svc, Associated with pulmonary atresia, pulmonary stenosis, Presence of IVC & abdominal aorta on same side– pathognomic. At chest radiography, the cardiac apex typically appears discordant from that of the stomach and liver. The stomach may also be midline and small (microgastria). In Peripheral smear- Look for howelljolly , heinz bodies and pitted erythrocytes. Electrocardiography shows Superior QRS axis, P axis is either normal (0 to +90 degrees) or alternating b/w the lower left and lower right quadrants. RVH, LVH, or BVH is present. The complex cardiac anomalies and abnormal immune status are closely linked poor prognosis of this group of patients. Death occurs in the first year of life in up to 80% of cases due to deficiencies of immunoglobulin Ig M&IgE, Impaired reticuloendothelial clearance and Depressed T cell function. Treatment options include PG infusion, Intra cardiac repair using modifications of fontan technique, Administration of Pneumococcal & H Influenza vaccine is recommended. Prognosis : poor, Mortality is as high as 90% Polysplenia is more common in females and has more variable clinical manifestations and prognosis. Classic left isomerism or bilateral left-sidedness implies that patients have bilateral bilobed lungs, bilateral pulmo-nary atria, a centrally located liver, a stomach in indeterminate position, and multiple spleens. Absence of the inferior vena cava on the lateral chest radiograph and the aygous continuation on the frontal chest radiograph with discordance of the apex and abdomin al viscera help suggest the diagnosis. In general, cardiac anomalies are less common in polysplenic patients and not as complex as those in asplenic patients. The most common cardiac anomalies in this group are partial anomalous pulmonary venous return, atrial septal defect, and atrioventricu-lar canal. Electrocardiography shows Ectopic atrial rhythm with a superiorly oriented P axis(-30 to -90 degrees). Superior qrs – endocardial cushion defects, RVH or LVH is common and Complete heart block-10%. Treatment: includes Surgical correction. Prognosis is better with low mortality. In summary, heterotaxy syndrome can be associated with numerous cardiac anomalies. Second trimester routine ultrasound evaluation of the fetal heart has been proposed for prenatal detection of cardiac anomalies. This report suggests that early intrauterine identification can accurately delineate the cardiac and vascular anatomy in heterotaxy, allowing planning prenatal counseling and postpartum planning for corrective surgery.

References:
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