Situs Inversus with Dextrocardia Associated with Complex Congenital Heart Disease: A Case Report

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Case Report

Abstract: Situs inversus is rare congenital anomaly, in which there is general transposition of viscera due to reverse rotation including heart which rotates to right side and all abdominal - thoracic viscera are laterally transposed i.e. all viscera normally on right are seen on left and vice versa. The heart is structurally normal in 90-95% of cases. Incidence of cardiac anomalies in dextrocardia with SI is low (3%), while in isolated dextrocardia it is high. Dextrocardia with situs inversus occurs in 2 per 10,000 live births. Establishment of body axis antero-posterior, dorso-ventral and right-left axis is during embryogenesis at the time of gastrulation.Gene expression patterns are responsible for establishing right-left axis, mainly hox code that specify segmental position. Recent research on mice suggests that situs inversus is caused by the absence of a single protein due to particular mutation on chromosome 12. We came across a 30 year old lady having this rare anomaly of situs inversus with dextrocardia associated with complex congenital heart disease.

Key words: situs inversus, dextrocardia, body axis.

Case History and Clinical Presentation

A 30 year old lady came with history of shortness of breath, palpitations and chest discomfort NYHA functional class II. Her general examination revealed signs of congestive heart failure, clubbing, central cyanosis and bilateral pitting pedal edema. On CVS examination, apex beat was in right 5th intercoastal space in anterior axillary line, loud S1 and pansystolic murmur. Other systemic findings were tender hepatomegaly and bibasal pulmonary crackles.

Investigations

ECG: S/O dextrocardia with Right Ventricular Hypertrophy (RVH).

X- Ray chest (P-A view) - Evidence of Dextrocardia with cardiomegaly with minimal pericardial effusion, gas in fundus of stomach on right side.

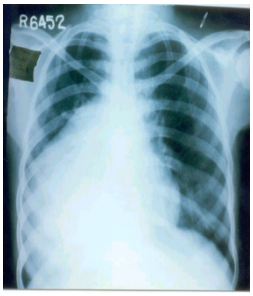

Figure 1: X- Ray Chest (P-A) : Dextrocardia, Cardiomegaly; Stomach fundus with gas on right side

USG Abdomen and Chest showed liver in left hypochondrium, dilated Inferior Vena Cava and hepatic veins, spleen in right hypochondrium, evidence of dextrocardia with pericardial effusion.

2-D ECHO and Color Doppler showed viscera-atrial situs inversus with dextrocardia, inversely related and dilated great arteries with pulmonary hypertension, dilated RA and RV cavities, large perimembranous ventricular septal defect with inlet extension with reversal of shunt, left sided normal aortic arch and normal biventricular functions.

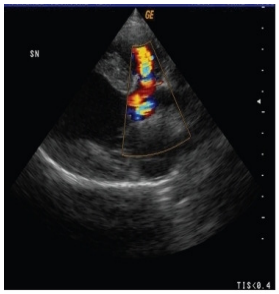

Figure 2: 2 D ECHO and COLOR DOPPLER: perimembranous ventricular septal defect with reversal of shunt

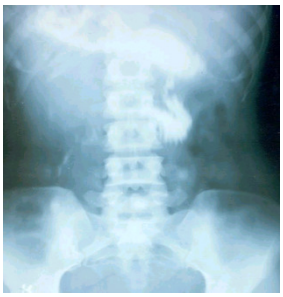

Figure 3: BARIUM MEAL showing stomach on right and "C" of duodenum on left side

Thus findings on clinical examination like apex beat in right 5th inter coastal space in anterior axillary line, X-ray, ECG, USG suggestive of liver in left hypochondrium, 2 D Echo with color Doppler transposition of great vessels, dextrocardia with VSD shows that patient has situs inversus with dextrocardia associated with VSD.

BARIUM MEAL FOLLOW THROUGH showed Stomach, Duodenum, Appendix and Caecum on right side suggestive of situs inversus viscerum.

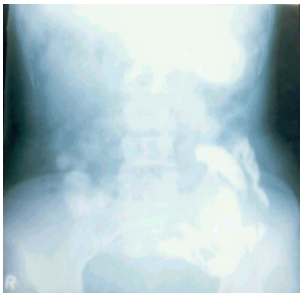

Figure 4: BMFT: Caecum and appendix on left side

Discussion

Situs inversus is a condition in which there is general transposition of the viscera due to reverse rotation including the heart which rotates to the right and all abdominal and thoracic viscera are laterally transposed i.e. all parts normally on the right are seen on the left and vice versa. Recent research in mice suggests that the situs inversus is caused by the absence of a single protein due to particular mutation on the chromosome 12 that is critical for the regulation of correct handedness. Dextrocardia with situs inversus occurs in approximately 2 per 10,000 live births. Incidence of cardiac defects is low among these individual- probably 3%. Dextrocardia with situs solitus or situs ambiguous is considerably less common- 1 in 20,000 live births in which incidence of CHD is extremely high- about 90% as compared to isolated dextrocardia and arterial transposition. The TGFB factor nodal is involved in looping of the heart tube. Its role in dextrocardia is unknown. Mutations in heart specifying gene nkx 2.5 result in heart defect.

VSD involving membranous portion of septum is the most common congenital cardiac malformation, about 80% defects are perimembranous or may extend into the inlet, trabecular, or outlet section of muscular septum although VSD may be isolated lesion, it is often associated with transposition of great vessels. Transposition of great vessels occurs when conotruncal septum fails to fall its spiral course and runs straight down, as a result aorta originates from the right ventricle to the right of and anterior to the pulmonary artery and pulmonary artery from the left ventricle. This results in two separate and parallel channels of circulation, and some communication between the two circulation must exist after birth to sustain life.Most patients have an interatrial communication, 2/3 have a PDA and VSD in the membranous part of interventricular septum. Incidence of VSD is 4.8/10,000 births.

Since neural crest cells migrating from the edges of the neural folds in the hindbrain region contribute to endocardial cushion formation in both the conus cordis and truncus arteriosus as well as interventricular septum formation, abnormal migration, proliferation or differentiation of these cells results in VSD, TGV, PDA etc. establishment of body axis antero-posterior dorso-ventraland the left right axis is established during embryogenesis at the time of gastrulation. The heart, associated vessels and inner organs adopt spatial arrangement and morphogenesis. Gene expression patterns are responsible for establishing left right axis. hox genes specify hox code that specify segmental position along various axis. Dorsal embryonic structure remain dorsal and undergo little change while originally midline structure and ventral structures especially derived from splanchno-pleuric mesenchyme such as CVS and gut are subject to extensive shifts.resulting in change in bilateral symmetric arrangement of a whole body. hox genes specify hox code that specify segmental position along various axis. hox gene expressed in developing ectodermal and mesenchymal structures such as neural crest somites, heart, kidney etc. but not in endodermaly derived structures; therefore error in interpretation of left right information result in congenital anomaly of heart, spleen, gut, major vessels.

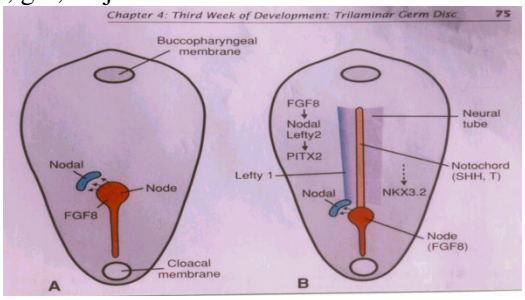

Figure 5: Gene expression patterns for establishing the left-right axis

Medical Management

Treatment of Congestive Heart failure: - Digoxin, Diuretics, Vasodilators, Oxygen, Rest, Salt restriction etc.

Advice

Surgical correction of VSD, TGV.

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