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Prenatal diagnosis of heterotaxy syndrome and autopsy correlation – a 12 year review of a single-centre experience

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Objective

To characterize the pathologic features of right and left isomerism in prenatally diagnosed cases of heterotaxy syndrome.

Methods

This was a retrospective case study conducted in a single tertiary level fetal medicine Centre over a period of twelve years from January 2004 to September 2016, with gestational age ranging from 15 to 29 weeks. Fetuses with ultrasound diagnosis of heterotaxy syndrome and also autopsy confirmation were included in the study. Heterotaxy was reported in prenatal ultrasound when there was abnormal thoracic or abdominal situs, detected with or without structural heart defects. Based on autopsy, the study group was subdivided into right isomerism or left isomerism. Right isomerism was defined based on presence of any of the following: 1) Bilateral right atrial appendages 2) Bilateral right sided lungs with or without asplenia. Left isomerism was defined based on any of the following criteria: 1) Presence of bilateral left atrial appendages 2) Bilateral left sided lungs with or without polysplenia. The frequency of distribution of various components of situs abnormalities and associated cardiac defects were analyzed.

Results

Over a period of twelve years there were 245 fetuses diagnosed antenatally with heterotaxy syndrome. Of these, 35 pregnancies were terminated and sent for autopsy and thus formed the study group. The median gestational age at diagnosis by ultrasound was 22 weeks. Of these 35 cases, 21 presented with levocardia, 12 cases had dextrocardia and 2 had mesocardia. Sixteen (16/35, 46%) cases had features of right isomerism and nineteen (19/35, 54%) had left isomerism, demonstrating an almost equal distribution of left and right isomerism in this series. The most common cardiac defect was atrioventricular septal defect (AVSD) seen in 27 cases (27/35, 77%). Of these, 13 were seen in right isomerism (48%) and 14 in left isomerism (52%). Double outlet right ventricle (DORV) was the most common outflow tract abnormality 12/35 (34%). DORV was seen more commonly in right isomerism (66%). Among the venous abnormalities, persistent left superior vena cava (LSVC) was most common (24/35, 68%), followed by anomalous pulmonary venous connection (APVC) in 43% (15/35). APVC was predominantly seen in right isomerism (80%), while persistent LSVC was equally distributed in both groups. Interrupted IVC was confirmed in 9 cases of which 8 were left isomerism. On prenatal ultrasound 92% (32/35) of the four chamber abnormalities were detected correctly. 3 cases reported as single ventricle on ultrasound, turned out to be AVSD in autopsy. Of these 3 cases, cardiac imaging was technically challenging for two cases and one presented at later gestation. DORV was diagnosed correctly in 8 out of 12 cases (66%) on prenatal ultrasound and the remaining 4 were diagnosed as outflow abnormality reported as anterior aorta and difficult imaging of pulmonary artery. 71% of LSVC and 53% of APVC were detected correctly on ultrasound. Interrupted IVC was diagnosed correctly in all the cases on prenatal imaging. Among the extra cardiac abnormalities intestinal malrotation was the most common, which was detected only by autopsy (15/35, 43%; 8 in left isomerism and 7 in right isomerism). Since all these 15 cases were imaged at early second trimester, malrotation has not been detected by ultrasound.

Conclusion

This large autopsy based series provided valuable insights into the prenatal presentation of situs abnormalities and the associated defects. Major cardiac defects are present in the vast majority and are amenable to a high degree of prenatal detection. Intestinal malrotation was the most common extracardiac manifestation which could not be suspected in second trimester imaging, but has significant implication on prenatal counseling.