

Prenatal sonographic diagnosis of Right Isomerism associated with Sliding Hiatal hernia

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Objective

We report a case of heterotaxy syndrome with right isomerism, complex congenital heart disease associated with hiatus hernia in a 19 weeks fetus. The objective of this report is to highlight the importance of having a proper checklist for prenatal identification of extracardiac manifestations of heterotaxy syndromes. This will enable us to provide an effective prenatal counseling and postnatal management of this complex lesions. To our knowledge, prenatal sonographic detection of a sliding hiatal hernia in a fetus with asplenia syndrome has not been reported previously.

Methods

A 42 years old G2P1L1 was referred at 19 weeks and 2 days for fetal cardiac evaluation in view of suspicion of congenital heart disease in an anomaly scan performed outside. Fetal echocardiogram performed using the Voluson E10 equipment (GE medical systems, Zipf, Austria) showed a complex congenital heart disease. (Right isomerism, situs ambiguous, dextrocardia, common atrium, common atrioventricular valve, large inlet and muscular ventricular septal defect amounting to a single ventricle, pulmonary atresia, total anomalous pulmonary venous connection, intact inferior vena cava and bilateral superior vena cava with sinus rhythm). Incidentally the stomach was seen in transverse section of fetal thorax at level of the four chamber view, hence congenital diaphragmatic hernia was suspected. The total lung area measured in four chamber view was within normal limits for the gestational age. The absence of mediastinal shift and normal appearance of the diaphragm pointed towards hiatus hernia. Fetal abdominal examination showed isolated complete herniation of the stomach. The complexity of sonographic and echocardiographic findings were explained to the couple and they opted for termination of pregnancy.

Results

A retrospective postnatal study showed a significant association of hiatus hernia in patients with right isomerism (14.3% n=17/143). The common clinical manifestations are vomiting, recurrent bronchiolitis or pneumonia and upper gastrointestinal bleeding. Patients with hiatus hernia can have associated midgut malrotation. There is an isolated case report on prenatal diagnosis of hiatus hernia (paraesophageal) associated with heterotaxy syndromes. In paraesophageal hiatus hernia, the gastroesophageal junction(GEJ) is confined to its normal position, but a part of the stomach herniates in to the chest. But in our case, the entire stomach including GEJ herniated in to the thoracic cavity. To our knowledge, prenatal sonographic detection of a sliding hiatal hernia in a fetus with asplenia syndrome has not been reported previously.

Conclusion

Right isomerism is typically associated with complex congenital heart malformations along with various extra cardiac manifestations. This case illustrates the importance of having a proper checklist for prenatal identification of extracardiac manifestations of heterotaxy syndromes. Fetal echocardiography along with detailed anomaly scan can accurately delineate the cardiac and extracardiac anatomy in heterotaxy syndromes, helping to plan effective prenatal counselling.