Prenatal diagnosis of asplenia syndrome with sliding hiatus hernia in a fetus

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ABSTRACT

We report a case of right isomerism with a complex congenital heart disease associated with hiatus hernia in a 19-week-old fetus with relevant review of literature. This report highlights the importance of having a proper checklist for prenatal identification of extracardiac manifestations of isomerism syndromes. This will enable us to provide an effective family-centered counseling for perinatal management of these complex lesions. To our knowledge, prenatal sonographic detection of a sliding hiatal hernia in a fetus with right isomerism has not been reported previously.

Keywords: Asplenia syndrome, fetus, sliding hernia

INTRODUCTION

Abnormal lateralization of the thoracic and abdominal organs, which are arranged in an unusual arrangement, is called isomerism syndrome.1 It is found in between 2.2% and 4.2% of infants with congenital heart disease (CHD). It is typically associated with malformations in the central nervous, pulmonary, gastrointestinal, immunologic, and genitourinary systems [Table 1].2,3 Previously, the syndrome was segregated on the basis of the anatomy of spleen into asplenia and polysplenia categories. However, it has been proven that the spleen is not the best discriminator between the two subsets of isomerism syndromes.4

CASE REPORT

A 42-year-old second gravida was referred to us at 19 weeks and 2 days for fetal cardiac evaluation in view of a suspicion of CHD on fetal anomaly scan. Fetal echocardiogram was performed using the Voluson E10 equipment (GE medical systems, Zipf, Austria), which showed a complex CHD bearing the diagnosis of right isomerism. The situs was ambiguous with dextrocardia [Figure 1]. Four-chamber view showed a common atrium, a common atrioventricular valve, a large inlet, and a muscular ventricular septal defect, amounting to a single ventricle [Figure 2]. The outflow tracts showed only a single outlet (aorta) with pulmonary atresia [Figure 3]. Venous system evaluation showed total anomalous pulmonary venous connection to the right superior vena cava with intact inferior vena cava and bilateral superior vena cava [Figure 4]. The cardiac rhythm was sinus with a fetal heart rate of 146 beats/ min.

In the four-chamber view, a hypoechoic structure was seen in the left fetal thoracic cavity, and the heart was visible in the right thoracic cavity in the transverse view [Figure 2]. The total lung area measured in four-chamber view was within normal limits for gestational age. There was no mediastinal shift, and the diaphragm had a normal appearance, thus excluding the possibility of a congenital...
diaphragmatic hernia. Fetal abdominal examination showed isolated complete herniation of the stomach and gastroesophageal junction (GEJ) [Figure 5]. Thus, the possibility of a sliding hiatus hernia was considered. An open neural tube defect in the occipital region suggestive of occipital meningomyelocele with kyphosis was noted [Figure 6]. Complexity of the CHD and the extracardiac abnormalities were explained to the parents, and they opted for medical termination of pregnancy.

Fetal autopsy was not performed because the parents refused for the same.

Table 1: Various extracardiac malformations associated with right isomerism

| CNS | Pulmonary | Gastrointestinal | Genitourinary | Immunologic |
|-----|-----------|------------------|---------------|-------------|
| Holoprosencephaly | Primary ciliary dyskinesia | Intestinal malrotation (70%) | Horseshoe kidney | Hypofuntion of spleen (asplenia [right isomerism]; polysplenia [left isomerism]) |
| Asymmetry in cerebral volumes | Right isomerism (bilateral trilobed lungs, bronchial angle>135°) | Hiatus hernia | Gastric volvulus | Ectopic ureters |
| Myelomeningocele | Left isomerism (bilateral bilobed lungs, bronchial angle<135°) | Gastric volvulus | Predudodenal portal vein | Ureteral duplication |
| Spina bifida | Omphalocele | Congenital diaphragmatic hernia | Tracheoesophageal fistula | Cystic kidneys |
| Cranioiacchisisis | Biliary atresia (left isomerism) | Omphalocele | Midline liver and gall bladder | Solitary kidney |
| Dandy-Walker syndrome | Agenesis of the dorsal pancreas | Omphalocele | Anus atresia | Cloacal duplication |
| Chiari II malformation | Anal atresia | Omphalocele | Portosystemic shunts | |
| Corpus callosum abnormalities | Omphalocele | Midline liver and gall bladder | Midline liver and gall bladder | |
| Aqueductal stenosis | Omphalocele | Omphalocele | Omphalocele | |
| Open neural tube defects | Omphalocele | Omphalocele | Omphalocele | |
| Spinal meningocoele | Omphalocele | Omphalocele | Omphalocele | |
| Occipital meningocoele | Omphalocele | Omphalocele | Omphalocele | |

Figure 1: (visceral situs): Midline liver, absent spleen, aorta to the right and inferior vena cava to right and anterior of aorta suggestive of right isomerism

Figure 2: Four-chamber view shows stomach on the left side and cardiac apex on the right side. Both stomach shadow and cardiac chambers are visible in the four chamber view

Figure 3: Three-vessel view showing bilateral superior vena cava and single outlet (dilated aorta) and absence of pulmonary artery suggesting pulmonary atresia. Asc-Ao- ascending aorta. Zs-GA: Z score for gestation age

Figure 4: A channel between left atrium and spine in the area behind the heart suggestive of total anomalous pulmonary venous return
DISCUSSION

Right isomerism is characterized by disruption of early left-right axis determination and bilateral right sidedness. It is typically associated with complex congenital heart malformations such as unbalanced complete atrioventricular septal defects along with asplenia and intestinal malformation. It generally carries a suboptimal prognosis. Distribution of cardiac and extracardiac anomalies is different in two types of isomerism syndromes [Table 2].

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. However, the overall mortality was similar between those patients with and without hiatus hernia. There was an isolated case report on the prenatal diagnosis of hiatus hernia (paraesophageal) associated with isomerism syndromes. In this report, the authors described a paraesophageal hiatus hernia, with the GEJ confined to its normal position with a part of the stomach herniating into the chest. However, in our case, the entire stomach including GEJ was herniating into 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

This case illustrates the importance of having a proper checklist for prenatal identification of extracardiac manifestations of isomerism syndromes. This will enable us to provide an effective family-centered counseling and perinatal management of these complex lesions.
Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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