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

Balaganesh Karmegaraj[1], Satjeet Kaur Deol[2], Vivek Krishnan[2], Balu Vaidyanathan[1]

[1]Departmentof Fetal Cardiology, Amrita School of Medicine, Amrita Institute of Medical Sciences, Kochi, Kerala, India, [2]Department of Fetal Medicine, Amrita School of Medicine, Amrita Institute of Medical Sciences, Kochi, Kerala, India

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 isomersim 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

Access this article online

Quick Response Code:

Website: www.annalspc.com

DOI: 10.4103/apc.APC_34_19

How to cite this article: Karmegaraj B, Deol SK, Krishnan V, Vaidyanathan B. Prenatal diagnosis of asplenia syndrome with sliding hiatus hernia in a fetus. Ann Pediatr Card 2020;13:167-70.

Address for correspondence: Prof. Balu Vaidyanathan, Department of Pediatric Cardiology, Fetal Cardiology Division, Amrita Institute of Medical Sciences, Kochi - 682 041, Kerala, India. E-mail: baluvaidyanathan@gmail.com

Submitted: 08-Mar-2019 **Revised:** 05-Jun-2019 **Accepted:** 05-Dec-2019 **Published:** 20-Mar-2020

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	Ectopic ureters	
Myelomeningocele	Left isomerism (bilateral bilobed lungs, bronchial angle<135°	Gastric volvulus	Ureteral duplication	
Spina bifida		Preduodenal portal vein	Cystic kidneys	
Craniotachischisis		Congenital diaphragmatic hernia	Solitary kidney	
Dandy–Walker syndrome		Tracheoesophageal fistula	Cloacal duplication	
Chiari II malformation		Omphalocele		
Corpus callosum abnormalities		Biliary atresia (left isomerism)		
Aqueductal stenosis		Duodenal atresia		
Open neural tube defects		Agenesis of the dorsal pancreas		
Spinal meningocele		Anal atresia		
Occipital meningocele		Portosystemic shunts		
		Midline liver and gall bladder		
		Annular pancreas		
		Cleft lip/palate		

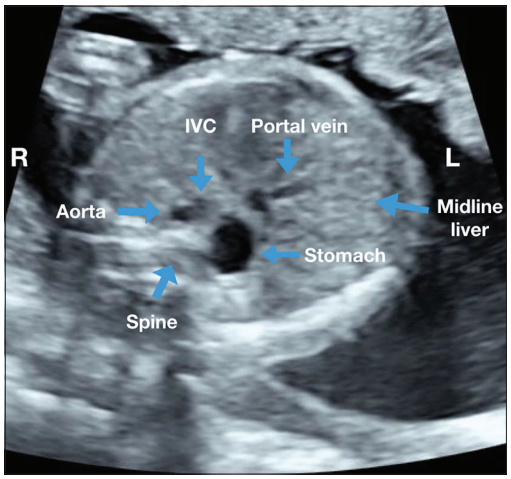

Figure 1: (visceral situs): Midline liver, absent spleen, aorta to the right and inferior vena cava to right and anterior of aorta suggestive of right isomerism

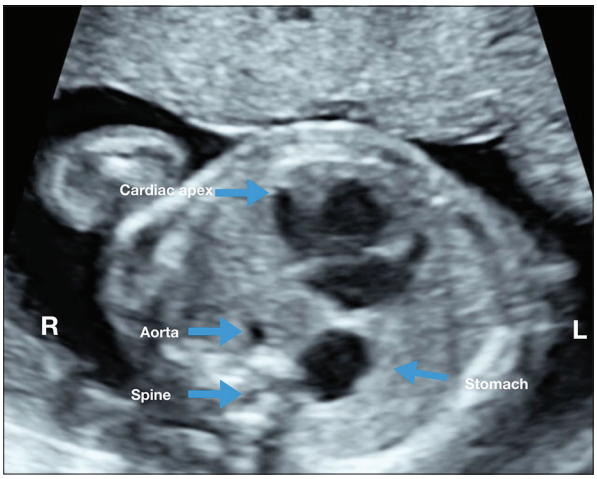

Figure 2: Four-chamber view shows stomach on the left side and cardiac apex on the right side. Both stomach shadow and cardiac chambers are visible in the four chamber view

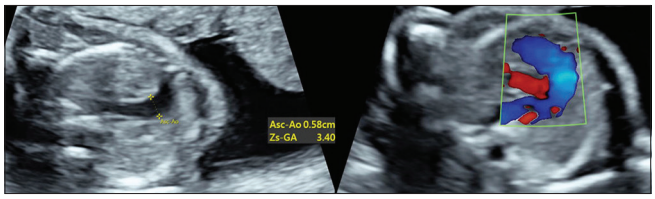

Figure 3: Three-vessel view showing bilateral superior vena cava and single outlet (dilated aorta) and absence of pulmonary artery suggesting pulmonary atresia. Asc-Ao- ascending aorta. Zs-GA: Z score for gestation age

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.

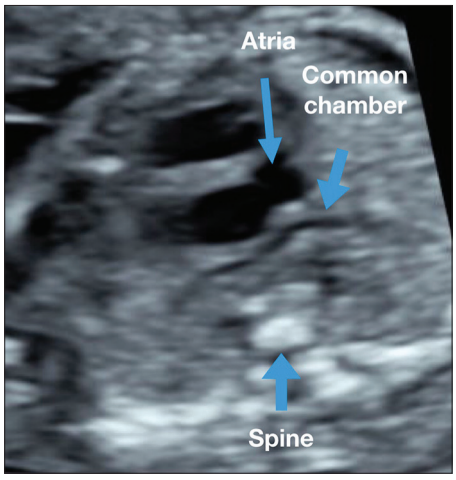

Figure 4: A channel between left atrium and spine in the area behind the heart suggestive of total anomalous pulmonary venous return

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

Table 2: Distribution of various cardiac and extracardiac malformations associated with right and left isomerisms

	Right isomerism (%)	Left isomerism (%)	Both
Extracardiac anomalies	Hiatus hernia Horseshoe kidney Encephalocele Achondroplasia	Biliary atresia Esophageal atresia Anal atresia Unilateral renal agenesis Dandy–Walker malformation Akinesia deformation sequence AV block Interrupted IVC (87%)	Situs ambiguus Dextrocardia Persistent LSVC Bowel atresia Volvulus Unilateral multicystic kidney Renal duplication
Cardiac anomalies (%)			
AV septal defect	67.9	71.4	
Univentricular physiology	24.5	11.6	
Right outflow obstruction	64.2	36.6	
Left outflow obstruction	13.2	11.6	
Double-outlet right ventricle	35.8	22.3	
Total anomalous pulmonary venous return	32.1	4.5	
Corrected transposition	28.3	8.9	
AV block	0	39.3	
Hydrops	9.4	29.5	
Any cardiac defect	100	86.6	

AV: Atrioventricular, IVC: Inferior vena cava, LSVC: Left superior vena cava

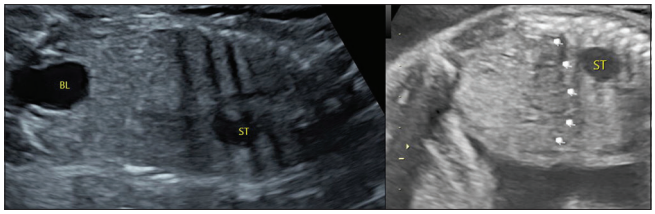

Figure 5: Coronal view showing complete herniation of the stomach including gastroesophageal junction in to the thoracic cavity with absence of stomach shadow in abdomen (finger mark showing the level of diaphragm). ST: Stomach: BL: Bladder

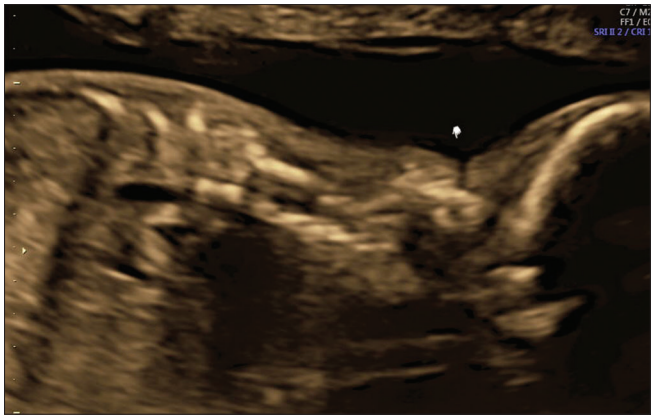

Figure 6: A neural tube defect in the occipital region suggestive of occipital meningomyelocele

DISCUSSION

Right isomerism is characterized by disruption of early left-right axis determination and bilateral right sidedness.[5] It is typically associated with complex congenital heart malformations such as unbalanced complete atrioventricular septal defects along with asplenia and intestinal malformation.[6] It generally carries a suboptimal prognosis.[7] Distribution of cardiac and extracardiac anomalies is different in two types of isomersim syndromes [Table 2].[8]

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.[9] 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.[10] 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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Jacobs JP, Anderson RH, Weinberg PM, Walters HL 3rd, Tchervenkov CI, Del Duca D, *et al.* The nomenclature, definition and classification of cardiac structures in the setting of heterotaxy. Cardiol Young 2007;17 Suppl 2:1-28.
2. Loomba R, Shah PH, Anderson RH. Fetal magnetic resonance imaging of malformations associated with heterotaxy. Cureus 2015;7:e269.
3. Kothari SS. Non-cardiac issues in patients with heterotaxy syndrome. Ann Pediatr Cardiol 2014;7:187-92.
4. Ivemark BI. Implications of agenesis of the spleen on the pathogenesis of conotruncus anomalies in childhood; an analysis of the heart malformations in the splenic agenesis syndrome, with fourteen new cases. Acta Paediatr Suppl 1955;44:7-110.
5. Hildreth V, Webb S, Chaudhry B, Peat JD, Phillips HM, Brown N, *et al.* Left cardiac isomerism in the Sonic hedgehog null mouse. J Anat 2009;214:894-904.
6. Anderson RH, Spicer DE, Loomba R. Is an Appreciation of Isomerism the Key to Unlocking the Mysteries of the Cardiac Findings in Heterotaxy? J Cardiovasc Dev Dis 2018;5. pii: E11.
7. Buca DIP, Khalil A, Rizzo G, Familiari A, Di Giovanni S, Liberati M, *et al.* Outcome of prenatally diagnosed fetal heterotaxy: Systematic review and meta-analysis. Ultrasound Obstet Gynecol 2018;51:323-30.
8. Gottschalk I, Stressig R, Ritgen J, Herberg U, Breuer J, Vorndamme A, *et al.* Extracardiac anomalies in prenatally diagnosed heterotaxy syndrome. Ultrasound Obstet Gynecol 2016;47:443-9.
9. Hsu JY, Chen SJ, Wang JK, Ni YH, Chang MH, Wu MH. Clinical implication of hiatal hernia in patients with right isomerism. Acta Paediatr 2005;94:1248-52.
10. Yamamoto N, Hidaka N, Anami A, Hojo S, Masumoto K, Taguchi T, *et al.* Prenatal sonographic diagnosis of a hiatal hernia in a fetus with asplenia syndrome. J Ultrasound Med 2007;26:1257-61.