Congenital lobar emphysema associated with polysplenia syndrome

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Polysplenia, or left isomerism, is a rare heterotaxy syndrome characterized by bilateral bi-lobed lungs, bilateral pulmonary atria, a symmetrical midline liver, and multiple aberrant splenic nodules. We report a case of polysplenia associated with congenital lobar emphysema apart from other typical anomalies. Such an association has not been previously reported. The patient was a young male with progressive exertional breathlessness referred for high resolution CT of the lungs. CT, MRI and echocardiography revealed (in addition to congenital lobar emphysema of right lung) a hemiazygos continuation of the inferior vena cava, a persistent left superior vena cava, multiple splenunculi in the right hypochondrium, midline liver, bilateral bilobed lungs, a large pulmonary artery (suggestive of severe pulmonary artery hypertension) and a large VSD—a typical constellation of findings described in polysplenia syndrome.

CASE
A 40-year-old man presented with insidious-onset progressive breathlessness. Physical examination showed mild central cyanosis, parasternal heave, a short systolic murmur, and a loud second heart sound. A chest radiograph revealed an enlarged pulmonary conus and hilar vessels and a radiolucent area in the right upper zone. High-resolution CT of the chest showed an enlarged main and lobar pulmonary arteries (suggestive of pulmonary arterial hypertension), with an expanded hyperlucent right upper lobe, suggestive of congenital lobar emphysema. The azygos and accessory hemiazygos veins were enlarged, with drainage of the hemiazygos into a persistent left superior vena cava (SVC), which drained into the coronary sinus (Figure 1). The liver was seen in the midline position with a short segment of infrahepatic IVC. The stomach was in the right upper quadrant with multiple splenic masses in relation to the greater curvature (Figure 2). MRI confirmed the presence of bilateral bilobed lungs with hyparterial bronchi and of the accessory hemiazygos continuation of the IVC (Figures 3, 4). Transthoracic and transesophageal echo revealed a large ventricular septal defect (VSD) with pulmonary arterial hypertension, which was suggestive of Eisenmenger syndrome. The patient was referred to a tertiary care cardiothoracic surgery department for further management. To the best of our knowledge this is the first reported case of polysplenia associated with congenital lobar emphysema.

DISCUSSION
The clinical manifestations in polysplenia vary and sometimes can be mild, with many patients (approximately 10%) surviving into mid-adolescence. No single abnormality is pathognomonic for polysplenia; hence, some authors prefer to use the term heterotaxy, and suggest that the different anatomical abnormalities be
The various cardiovascular anomalies that may be encountered include atrial septal defect (ASD) (78%), VSD (63%), partial anomalous pulmonary venous return (39%), transposition of the great arteries (31%), right-sided aortic arch (44%), pulmonary valvular stenosis (23%), and subaortic stenosis (8%). In addition, azygous continuation of the IVC is seen in 65% of cases and bilateral SVC in approximately 47%.1,3

The abdominal findings may include midline liver (57%), situs inversus (21%), short pancreas, semi-annular pancreas, and predouenal portal vein.4–7 Renal agenesis or hypoplasia may also be seen.4–7 Malrotation, including nonrotation, reverse rotation, and midgut volvulus, is also frequently seen in heterotaxy.5,9 CT as well as MRI can be used to characterize both the visceral and the cardiovascular anomalies in polysplenia and can provide necessary information for surgical planning.10 The antenatal diagnosis of cardiopulmonary syndromes is possible by sonography supple-
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The pulmonary manifestations include bilateral bilobed lungs and hyparterial bronchi (58%).1,2 Our patient had a hyperlucent upper lobe with attenuated vascular markings, which was suggestive of congenital lobar emphysema, an entity not described in polysplenia until now. However, the association of congenital heart disease (patent ductus arteriosus, atrial septal defect, ventricular septal defect, total anomalous pulmonary venous return, Tetrology of Fallot) with congenital lobar emphysema is well known12,13 and is reported to occur in 14% to 50% of cases. Both ultrasound and MRI are useful in the antenatal diagnosis of congenital lobar emphysema as well as other bronchopulmonary malformations. Ultrasound depicts congenital lobar emphysema as a distended fluid-filled anechoic mass that may decrease in size as the gestation progresses. Fetal MRI reveals the high-signal expanded lobe, with compression of the remaining lung and mediastinal deviation (on T2* GRE sequences).14,15 In our patient, congenital lobar emphysema must have contributed to the worsening of the pulmonary arterial hypertension and dyspnea. The discovery of a bronchopulmonary malformation, including congenital lobar emphysema, on routine antenatal ultrasound should be followed by dedicated fetal echocardiography and a specific search for viscerocardiac heterotaxy.

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