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*Brief Report*

# Septum primum malposition defect and inferior sinus venosus defect: a rare association

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**Abstract** We present a case of a 2-year-old girl who presented with respiratory distress and umbilical abscess, and was found to have an inferior sinus venosus defect, malposition of the atrial septum primum, absent septum secundum, and anomalous drainage of the right upper and lower pulmonary veins to the right atrium.

Keywords: Inferior sinus venosus defect; septum primum malposition defect; partial anomalous pulmonary venous drainage

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## History

A 2-year-old girl presented to our institution after a recent umbilical hernia repair for respiratory distress and an umbilical abscess. The patient's vital signs revealed a heart rate of 181 beats/minute, respiratory rate of 80, and oxygen saturation of 90% on room air that improved to 95% on 3 L of oxygen via the nasal cannula. Owing to difficulty in weaning her off oxygen, a chest X-ray was performed that demonstrated mild cardiomegaly, right atrial enlargement, and pulmonary interstitial oedema. These findings prompted a cardiac evaluation, which identified a grade 1/6 low-pitched systolic murmur at the upper left sternal border. Her electrocardiogram showed peaked P-waves consistent with right atrial enlargement and tall R (20 mm)-waves in V1, consistent with right ventricular hypertrophy. The echocardiogram demonstrated severe right atrial dilatation, malposition of the septum primum to the left, inferior sinus venosus defect, and possible anomalous drainage of all of the right-sided pulmonary veins to the right atrium (Fig 1a–d).

Because of this unusual anatomy and to better define the anomalous pulmonary venous drainage, a cardiac CT scan was performed, which confirmed the absence of the septum secundum, inferior sinus venosus defect, and septum primum malposition defect. In addition, it further delineated the pulmonary venous anatomy, demonstrating anomalous drainage of the right upper pulmonary vein to the right atrium and the right lower pulmonary vein at the level of inferior sinus venosus defect, having normal connection to the left atrium (Fig 2a–e). We have elected to allow our patient to grow and recover from her illness before referring for surgical repair.

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## Discussion

We report a rare case of inferior sinus venosus defect associated with the absent septum secundum and septum primum malposition defect, resulting in an anomalous venous drainage of the right upper and lower pulmonary veins to the right atrium, with normal pulmonary venous connection to the left atrium. Inferior sinus venosus defect and septum primum malposition defects are both rare cardiac malformations and their association is extremely rare and only two cases have been reported to date.[1]

Inferior sinus venosus defect is a rare veno-atrial communication located posteriorly and inferiorly in the mouth of the inferior caval vein along with anomalous right pulmonary venous drainage to the inferior caval vein, while the pulmonary veins retaining normal connection to the left atrium.[2,5] It accounts for about 3% of atrial septal defects[3] and 5% of sinus venosus defects.[4]

In one study, it was found that 50% of the patients with inferior sinus venosus defect were misdiagnosed

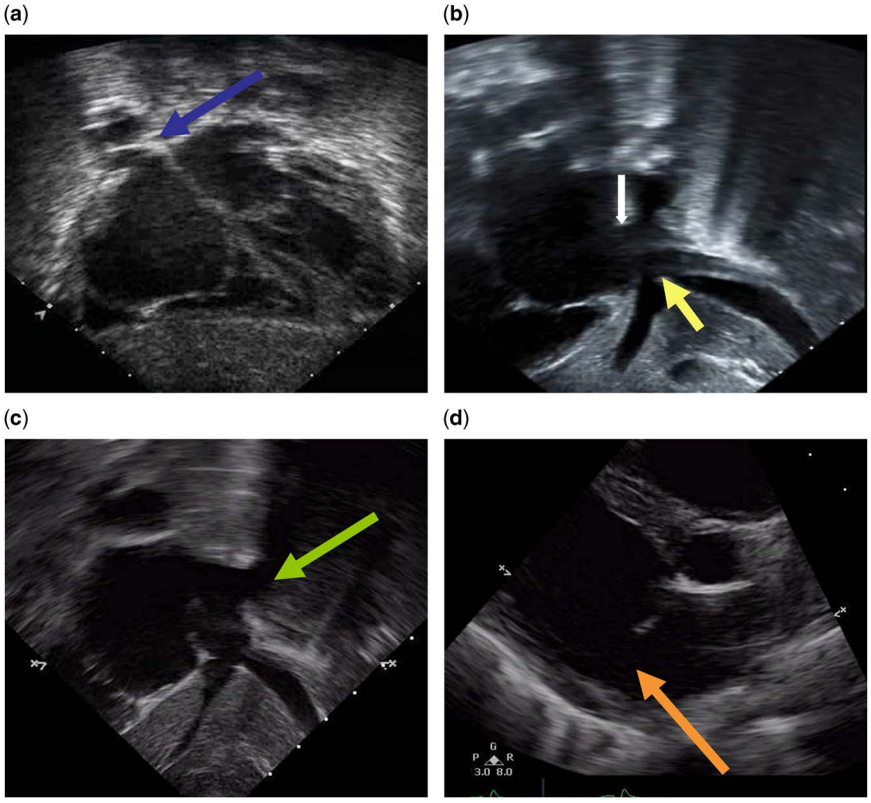


**Figure 1.**
*(**a**) Sub-xiphoid long-axis view demonstrating septum primum malposition defect and anomalous drainage of the right upper pulmonary vein (blue arrow). (**b**) Sub-xiphoid short-axis view demonstrating overriding of the inferior caval vein (yellow arrow) and the absence of the septal tissue at the mouth of the inferior caval vein. The atrial septum is marked by a white arrow. (**c**) Sub-xiphoid sagittal view illustrating the anomalous drainage of the right lower pulmonary vein (green arrow). (**d**) Parasternal short-axis view illustrating a defect in the posteroinferior part of the atrial septum (orange arrow).*

as secundum atrial septal defect by echocardiography preoperatively, but the authors found that the surgical outcomes were not affected by this misdiagnosis.[5] Another study comparing the outcomes of secundum atrial septal defect and inferior sinus venosus defect found that 64% of the patients with inferior sinus venosus defects were misdiagnosed by preoperative echo imaging and 40% intraoperatively, but the authors demonstrated preoperative diagnostic accuracy improving over time.[6] This study had disparate outcomes from the first, demonstrating worse surgical outcome, prolonged cardiopulmonary bypass time, longer ICU stay, and increased rate of reintervention as compared with the secundum atrial septal defect group.[6]

Accurate preoperative diagnosis is important to improve the perioperative outcome and decrease the incidence of reintervention for residual defects. With the current technological advancement, transthoracic echocardiography can accurately diagnose the defect in majority of children with sub-xyphoid and parasternal short-axis views, demonstrating overriding of the inferior caval vein with the absence of the septal tissue at the mouth of the inferior caval vein;[2,5,7] however, it may be difficult in obese and older patients because of poor sub-xyphoid windows. In such patients, alternative imaging modalities such as transoesophageal echocardiogram, cardiac CT, or cardiac MRI can be helpful to clearly delineate the defect and coexisting anomalous pulmonary venous drainage.[7–10] Cardiac MRI has an additional benefit of volumetric analysis and shunt quantification using phase-contrast flow measurements.[8]

Excellent surgical results have been reported utilising various surgical approaches, including closure of the inferior sinus venosus defect by displacement of the posterior edge of the atrial septum and suturing it to the right of the right pulmonary veins[11] and reimplantation of the anomalous pulmonary vein along with direct suture closure of the defect,[12] closure of the defect with a patch or construction of a baffle to redirect the pulmonary venous flow to the left atrium.[5]

Septum primum malposition defects have been speculated to result from displacement of the septum primum as a result of underdevelopment of the

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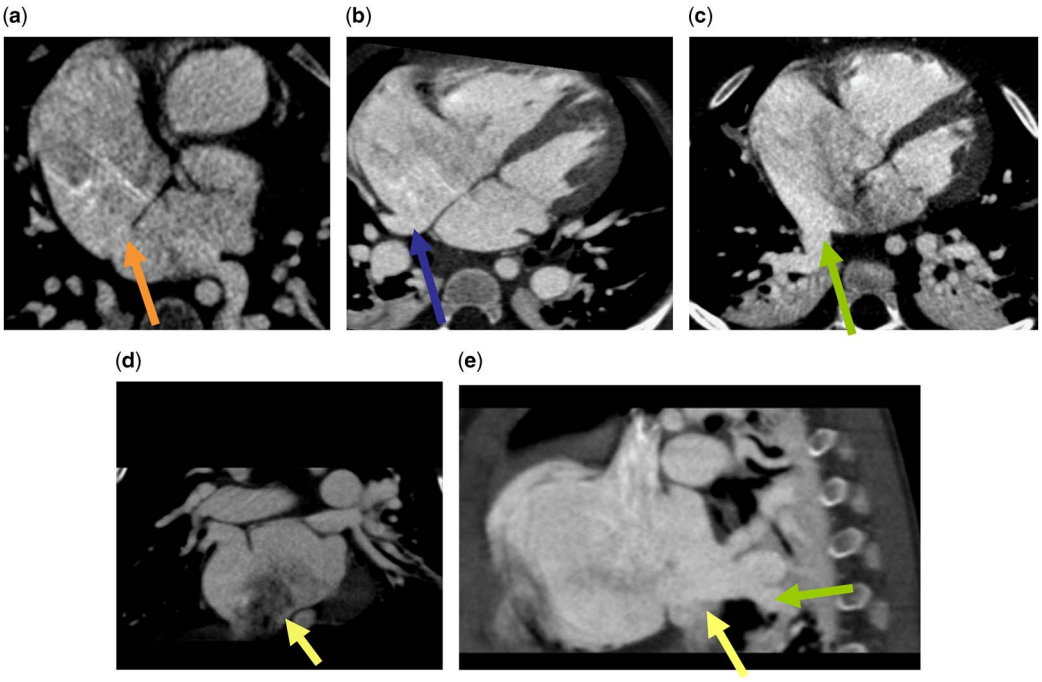


**Figure 2.**
*(**a**) Cardiac CT demonstrating a defect in the posteroinferior part of the atrial septum (orange arrow). (**b**) Cardiac CT illustrating septum primum malposition and anomalous drainage of the right upper pulmonary vein to the right atrium (blue arrow). (**c**) Cardiac CT illustrating septum primum malposition and the anomalous right lower pulmonary vein (green arrow) to the right atrium. (**d**) Cardiac CT demonstrating overriding of the inferior caval vein and a large defect at the mouth of inferior caval vein. (**e**) Cardiac CT illustrating the anomalous drainage of the right lower pulmonary vein into the right atrium (green arrow), just above the entrance of the inferior caval vein (yellow arrow).*

septum secundum, which allows anomalous drainage of half or all of the pulmonary veins to the right atrium, but having normal connection to the left atrium.[13] It has been shown to be associated with visceral heterotaxy in majority of patients, of which 92% had polysplenia and 87% had rhythm disturbances.[13] Our patient is unusual in that she had neither of these findings. In one study, it was demonstrated that one was able to accurately delineate the defect with two-dimensional echocardiography with sub-xyphoid coronal, parasternal short-axis, and apical four-chamber views.[14] Anatomic repair was achieved by atrial septal incision at the posterior edge and repositioning to redirect pulmonary venous flow to the left atrium and excellent results have been reported.[15]

## Conclusion

Septum primum malposition defects and inferior sinus venosus defect are rare cardiac malformations, which are associated with anomalous pulmonary venous drainage to the right atrium and normally connected to the left atrium. To date, the association of septum primum malposition defect with inferior sinus venosus defect has been reported only in two cases. Transthoracic echocardiography is an excellent non-invasive imaging modality to accurately diagnose both defects and associated anomalous pulmonary venous drainage in a majority of patients. We recommend additional imaging, such as cardiac CT or MRI in patients with doubtful diagnosis and to clearly delineate pulmonary venous anatomy. A high index of suspicion is essential for accurate preoperative diagnosis, which can significantly improve perioperative outcome and reduce the need for reintervention.

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## Conflicts of Interest

None.

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