

Case report

Prenatal diagnosis of a unique scimitar syndrome variant – a case report



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Abstract

Scimitar syndrome is a rare condition characterised by abnormal pulmonary venous return to the inferior vena cava. It is associated with various cardiopulmonary malformations such as atrial-septal defects, aortopulmonary collateral vessels, and pulmonary hypoplasia. Diagnosis is typically within the first few months of life. We report a unique case of scimitar syndrome that was diagnosed prenatally using multiple imaging modalities. In addition to two right-sided scimitar veins draining into the suprahepatic inferior vena cava, this patient was found to have horseshoe lung morphology with right pulmonary hypoplasia, a perimembranous ventricular septal defect, sequestered lung tissue, and an aortopulmonary collateral vessel arising from the descending aorta. The prenatal diagnosis allowed for timely medical and surgical intervention in the postnatal period, which is detailed in this report.

Key words: scimitar syndrome, foetal imaging, horseshoe lung, dextrocardia, sequestered lung.

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Introduction

Scimitar syndrome is a constellation of cardiopulmonary malformations characterised by the presence of anomalous pulmonary venous return to the inferior vena cava (IVC) from the right pulmonary vein (RPV). Commonly associated malformations include right-sided lung and pulmonary artery hypoplasia, sequestered lung tissue, aortopulmonary collateral vessels, horseshoe lung, and atrial septal defects [1]. It is estimated to affect 2/100,000 live births and has a 2:1 female-to-male predominance. There are two phenotypic variants of this syndrome: infantile and isolated. The infantile variant is strongly associated with additional malformations and presents symptomatically in early life, while the latter is often asymptomatic into adulthood and is characterised by isolated anomalous ve-

nous return [2–4]. The infantile variant is typically diagnosed within the first few months of life. Prenatal diagnosis is uncommon and has scarce mention in the literature. Visualisation of certain malformations associated with scimitar syndrome is possible, although comprehensive prenatal diagnosis through foetal imaging remains relatively undescribed [4–6]. This case illustrates the utilisation of multi-imaging modalities for the prenatal diagnosis of a rare presentation of scimitar syndrome and its postnatal medical and surgical management.

Case report

A 35 y/o G2P1 at 35 1/7 weeks was referred to paediatric cardiology for an abnormal obstetrical ultrasound, which showed rightward displacement of the heart into the right hemi-

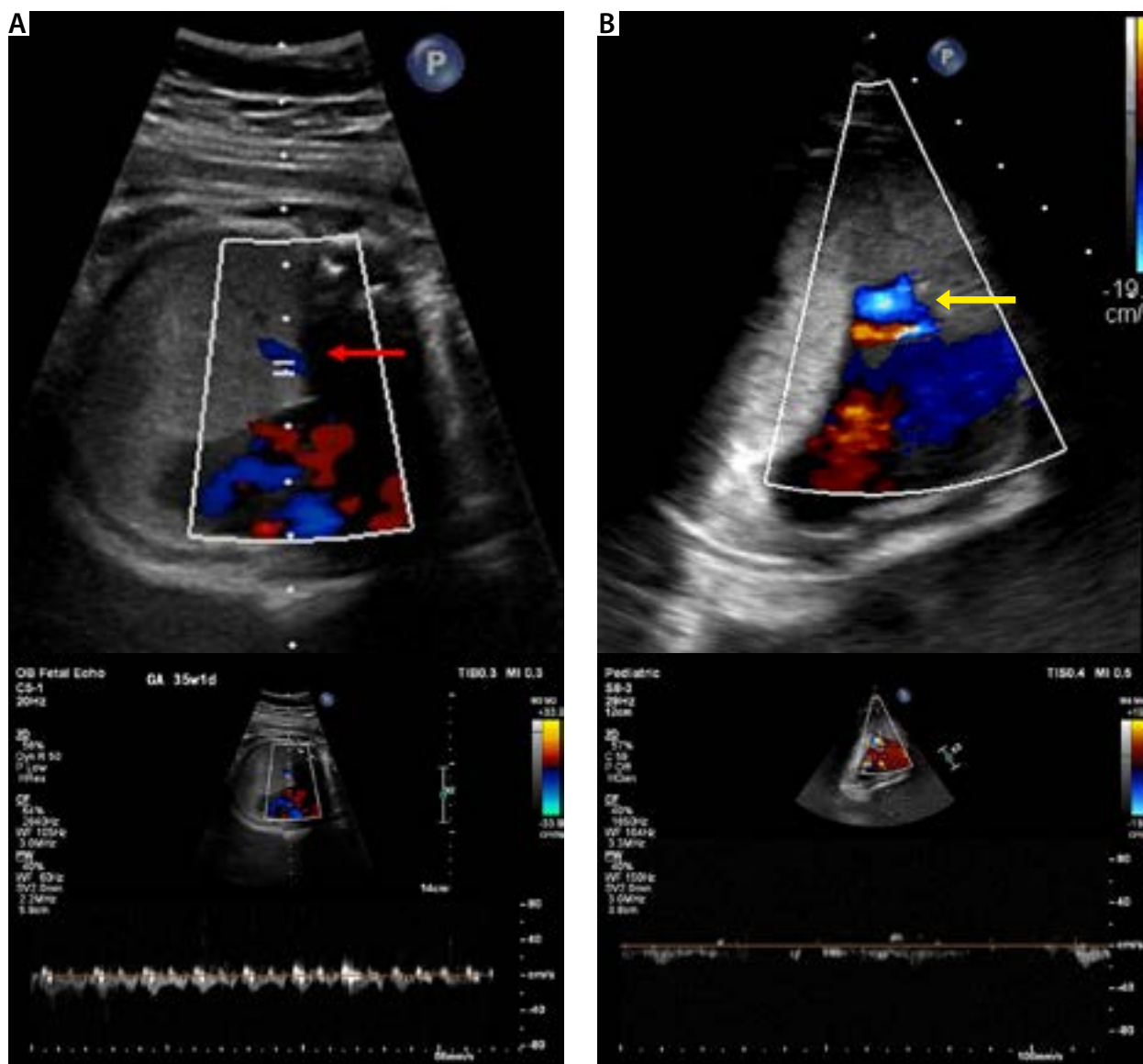


Figure 1. A – Foetal echocardiography with corresponding spectral Doppler demonstrating prominent drainage of the left pulmonary vein to the left atrium (red arrow) and B – abnormal venous return from the right pulmonary vein (yellow arrow). Significant dextroposition also noted. See supplemental videos “Foetal Echo RPV” and “Foetal Echo LPV”

thorax with suspected ventricular septal defect (VSD). Amniocentesis demonstrated 46XX karyotype with normal microarray. The prior pregnancy was uncomplicated, and there was no family history of congenital heart disease. Foetal echocardiography confirmed the presence of a moderate perimembranous VSD and dextroposition with levocardia. Pulmonary arterial anatomy and venous return on the left appeared normal. However, the right pulmonary artery was hypoplastic and right sided pulmonary venous return could not be visualised (Figure 1, Supplementary Figures S1, S2). The patient was then referred for foetal magnetic resonance imaging (MRI) to evaluate for possible anomalous pulmonary venous return and other underlying causes of dextroposition with mediastinal shift. Multiplanar, non-contrast MRI showed normal left-sided pulmonary anatomy with a truncated right mainstem bronchus and hypoplastic right lung. A vertical flow void from the midportion of the right lung was visualised, at which point scimitar syndrome

became the leading diagnosis. An additional flow void at the superior aspect of the descending aorta was detected, indicating a possible aortopulmonary collateral (Figure 2).

Echocardiography at birth showed partial anomalous RPV return to the IVC, moderate secundum atrial septal defect, large perimembranous VSD, and small patent ductus arteriosus. Cardiac CT showed two scimitar veins draining into the intrahepatic IVC with a single RPV draining the medial segment of the right lung to the left atrium. The RPA was markedly hypoplastic with atypical trifurcation with lower branch coursing left of the midline. CT also showed “horseshoe lung” morphology with incomplete pleural separation of the right and left lungs with the right lung lobe extending posteriorly into the left hemithorax (Figure 3). There was also a sequestration in the posterior right lung base with an aortopulmonary collateral arising from the descending aorta (Figure 4, Supplementary Figure S3).

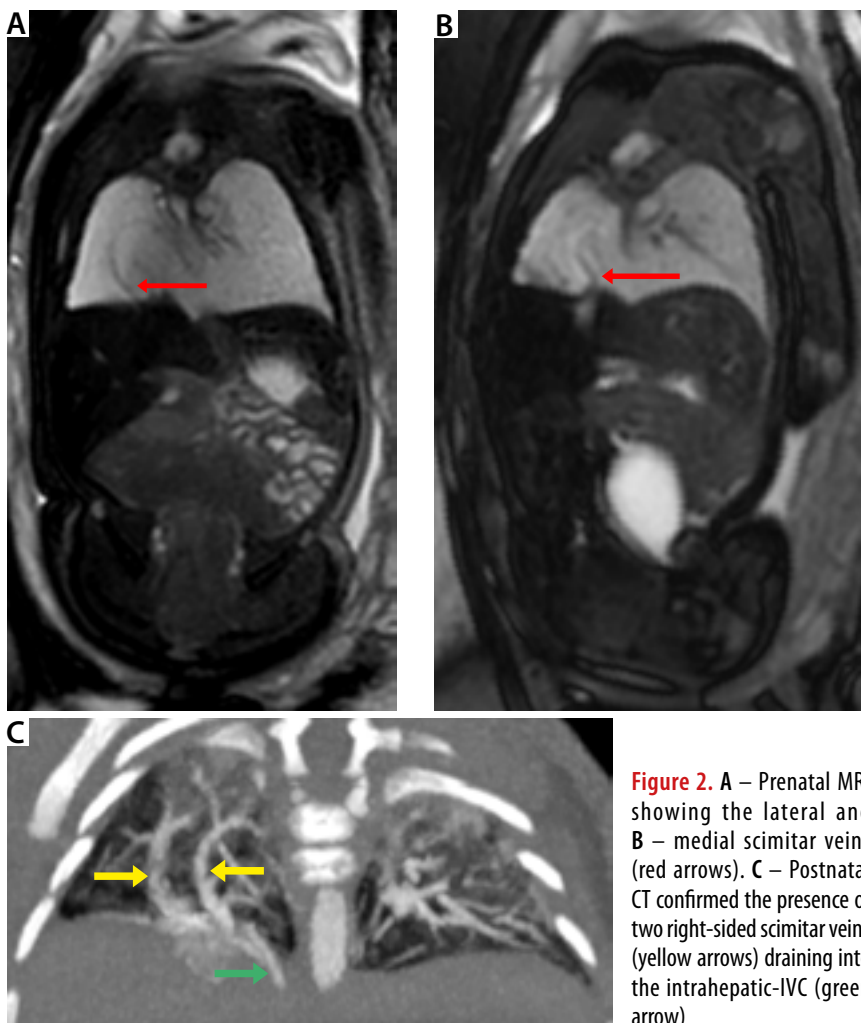


Figure 2. A – Prenatal MRI showing the lateral and B – medial scimitar veins (red arrows). C – Postnatal CT confirmed the presence of two right-sided scimitar veins (yellow arrows) draining into the intrahepatic-IVC (green arrow)

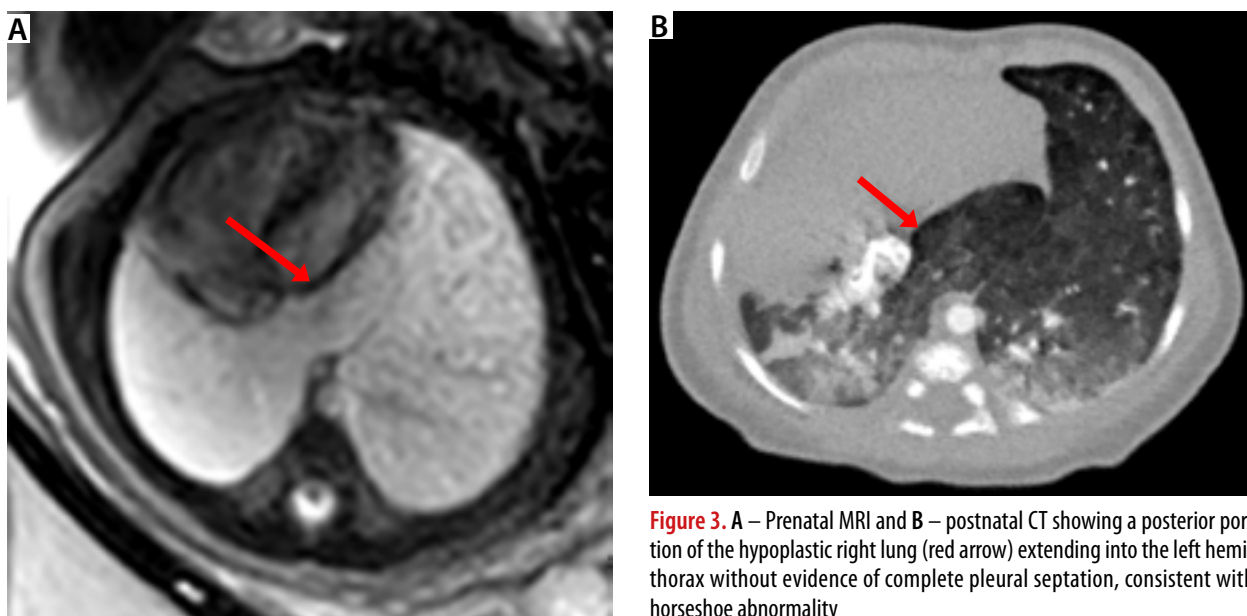


Figure 3. A – Prenatal MRI and B – postnatal CT showing a posterior portion of the hypoplastic right lung (red arrow) extending into the left hemithorax without evidence of complete pleural septation, consistent with horseshoe abnormality

Postnatally, the patient demonstrated baseline tachypnoea and retractions at baseline that worsened with feeding. The decision was then made to embolise the aortopulmonary vessel during cardiac catheterisation in order to improve the patient’s

cardiopulmonary physiology. The Qp:Qs ratio was 1.8:1 with LPA mean gradient of 24 mm Hg. Despite the use of furosemide and captopril, the patient continued to show evidence of congestive heart failure. She underwent surgical closure of the

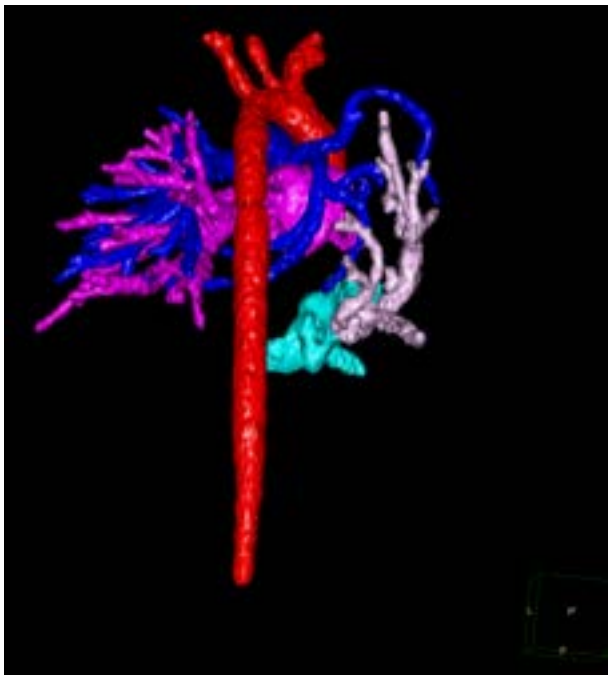


Figure 4. Posterior midline view of 3D image reconstructed from CT imaging. The hypoplastic right pulmonary artery (dark blue) supplies the hypoplastic lung, which is drained by two scimitar veins (grey) draining into a portion of the suprahepatic IVC (teal). Normal left pulmonary venous drainage to the left atrium (pink). See supplemental video “3D reconstructed image”

VSD, partial ASD closure, and PDA ligation. The postoperative course was complicated by pulmonary hypertension and sildenafil was started. Tachypnoea and retractions resolved by two months postoperatively without evidence of elevated pulmonary artery pressure by echocardiography, and sildenafil was discontinued. Over the next several months she demonstrated good weight gain with normal oxygen saturations. She has not subsequently had any significant respiratory infections or readmissions to the hospital. She is currently 1.5 years old without evidence of significant developmental delay or limitations in activity tolerance. The most recent echograph shows no evidence of intracardiac shunting, mildly dilated and hypertrophied right ventricle, normal biventricular systolic function, and scimitar venous drainage to the IVC.

Discussion

This case report describes the utilisation of various imaging modalities to establish the prenatal diagnosis of a unique variant of scimitar syndrome. In our review of the literature, we found only one case series detailing prenatal diagnosis. Bhide et al. showed that visualisation of abnormal pulmonary venous return was possible in four of six patients using a slow transverse sweep from the level of the four-chamber view down to the upper abdomen, closely examining the area surrounding the cavoatrial junction [7]. Although the scimitar vein could not be visualised by echocardiography, the lack of normal right-sided pulmonary venous return and associated mediastinal shift was worrying, because right lung hypoplasia is present in 90-100% of cases of scimitar syndrome and frequently

presents with dextroposition [2, 4]. This prompted advanced prenatal imaging providing the prenatal diagnosis of scimitar syndrome. Additionally, a flow void detected at the superior portion of the descending aorta raised suspicion for an aortopulmonary collateral. Detection of aortopulmonary collaterals to sequestered lung using foetal echocardiography is challenging because there is minimal flow through these vessels in the foetal period [6, 7]. This case demonstrates that foetal MRI is useful in visualising the scimitar veins as well as assessing for aortic supply to the sequestered lung.

The management of infantile scimitar syndrome includes various medical and surgical approaches tailored to each patient’s unique cardiopulmonary physiology. While definitive surgical repair can be performed after appropriate weight gain, the initial management should be directed by medical and catheter-based interventions [2]. The development of pulmonary arterial hypertension (PAH) is associated with higher mortality and is an important prognostic factor at the time of diagnosis [5]. Ligation or coil occlusion of aortopulmonary collaterals is an important early intervention; it has been demonstrated to reduce pulmonary artery pressure through reduction of haemodynamic load delivered to the anomalously drained sequestered lung tissue. This same principle can be applied to the correction of congenital heart disease contributing to pulmonary overcirculation [8, 9]. Furthermore, the presence of unrepaired congenital heart disease is associated with mortality and poor outcomes in patients who undergo definitive surgical repair of scimitar veins and those who are managed conservatively [4, 5]. In this case, embolisation of the aortopulmonary collateral did not provide sufficient reduction in haemodynamic load; she continued to demonstrate signs of high-output heart failure. Following correction of her underlying cardiac malformations she began to gain weight and has shown no signs of heart failure, albeit after a short postoperative course of PAH. Elective surgical repair of the scimitar veins is a possibility in the patient’s future, although it has been shown to only benefit infants who have a Qp:Qs ratio greater than 2. The majority of cases have low volume venous return from the hypoplastic lung and do not benefit from corrective surgery [5, 9]. Although our patient has responded well to the current catheter and surgical interventions, she has duplicate scimitar veins, suggesting that she may develop significant pulmonary overcirculation in the future.

Scimitar syndrome includes a spectrum of possible cardiopulmonary abnormalities. The embryologic pathogenesis is poorly understood, although it has been suggested that it is a result of abnormal development of blood supply and drainage of the developing lung bud. A plexus derived from the descending aorta initially supplies the lung buds, with a subsequent transition in the seventh week of life, in which the embryologic precursor of the pulmonary artery proliferates and becomes the main vascular supply. The umbilico-vitelline and cardinal veins initially drain the lung buds via the splanchnic plexus, which involutes as the plexus forms the common pulmonary vein along with the developing posterior left atrial wall. Insult

to this development perhaps explains the classical findings associated with this syndrome [2]. The association of horseshoe lung with scimitar syndrome is not well understood. While it is not as common as other malformations, the majority of reported horseshoe lung cases are found in infants with scimitar syndrome [10]. The communication of pleural spaces is a result of the failure of splanchnic mesoderm to septate, which normally occurs around the third week of life. It is suggested that improper pleural septation is associated with underlying structural heart defects because the timing parallels the septation of the cardiac tube. While horseshoe lung is usually not a direct cause of hypoxaemia or respiratory distress, the finding is clinically significant because incomplete septation of the pleura allows for contralateral extension of pneumothoraces [10, 11]. Up to 70% of scimitar syndrome cases are associated with ASDs, while additional structural defects are found in 20–30% of cases, the most common of which are VSDs and PDAs [4, 5]. Variants of classical scimitar syndrome have been described in which infants have some but not all of the trademark features. The “left-sided variant” mirrors classical scimitar syndrome, with an anomalous left scimitar vein draining into the IVC or azygous vein [1].

Conclusions

Scimitar syndrome is a rare condition associated with various cardiopulmonary malformations. This case has certain unique features that might share common embryological pathogenesis relating to the developing lung bud and its blood supply. While it is typically diagnosed in the first year of life, we demonstrated successful prenatal diagnosis of a rare variant of scimitar syndrome with associated horseshoe lung using various imaging modalities. Early diagnosis could lead to better outcomes because early intervention, particularly the embolisation of aortopulmonary collaterals, is associated with reduction in the risk of developing PAH.

Conflict of interest

The authors declare no conflict of interest.

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