Letter to the Editor

Prenatal diagnosis of a left superior vena cava draining into the left atrium and atrial septal defect with ventricular septal defect

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Introduction

Raghib syndrome is an infrequent developmental syndrome. It consists of the draining of a left superior vena cava (LSVC) into the left atrium (LA), the lack of the coronary sinus (CS), and an atrial septal defect (ASD) at the posterior-inferior angle of the atrial septum. Clinically, the increased pulmonary blood flow and cyanosis are characteristic malformations of Raghib syndrome (1). Further serious complications, such as chronic hypoxemia, infection, necrotizing fasciitis, and brain abscess have been reported (2,3). Consequently, prenatal diagnosis of this syndrome is recommended. Both prenatal and postnatal ultrasound diagnosis of this disease is very difficult and only a few cases have been reported in fetus. We present here a rare case of a LSVC draining into the LA and ASD located at the posterior-inferior angle of the atrial septum. This condition was diagnosed prenatally by echocardiography.

Case presentation

A pregnant woman, 25-year-old, gravida 2, was referred at 31+4 weeks of gestation as suspected ventricular septal defect was suspected. Amniocentesis performed at 20 weeks' gestation showed a normal karyotype. Routine fetal sonographic screening showed an abnormal four-chamber view. A moderate muscular ventricular septal defect was observed (*Figure 1A*) and the atrial septum appeared normal. Color Doppler recordings showed bidirectional flow within the defect (*Figure 1B*). To the left of the ductal arch a LSVC was detected in the three-vessel view (*Figure 1C*). The left longitudinal view showed that the LSVC drained into the LA with no bridging vein to the right superior vena cava (*Figure 1D*,*1E*). The opening of the LSVC in LA was in the left superior aspect next to the left upper pulmonary vein (*Figure 1F*). There was also a wide communication between two atriums with bidirectional flow. We could not detect a valve of foramen ovale in the interatrial position (*Figure 1D*). There was no septal tissue near the inferior vena cava (IVC). The diagnosis of ASD was made. The defect was located postero-inferiorly near the ostium of the IVC, which drained into the RA. A detailed examination of the heart did not show a CS. The two superior vena cava were approximately equal in size. The cardiac chambers were normal in size. Echocardiography revealed no valvular regurgitation.

A follow-up examination revealed no signs of cardiac dilatation or heart failure. An ultrasonic cardiogram, amniotic fluid volume, fetal growth, and Doppler flow waveforms of the middle cerebral artery, umbilical artery, IVC and ductus venosus were normal.

A female neonate weighing 2,800 g was delivered vaginally at 39+5 weeks' gestation following spontaneous labor. An examination showed that she was mildly cyanotic, with no dyspnea. Her oxygen saturation was 88–92% in air and 90–97% in 25% oxygen.

The prenatal findings were confirmed on postnatal assessment. Echocardiography showed the prenatally suspected muscular ventricular septal defect (diameter: 4.0 mm) with bidirectional flow (*Figure 2A*,*2B*). A LSVC was observed to flow directly into the upper left corner of LA between the left upper pulmonary vein and the

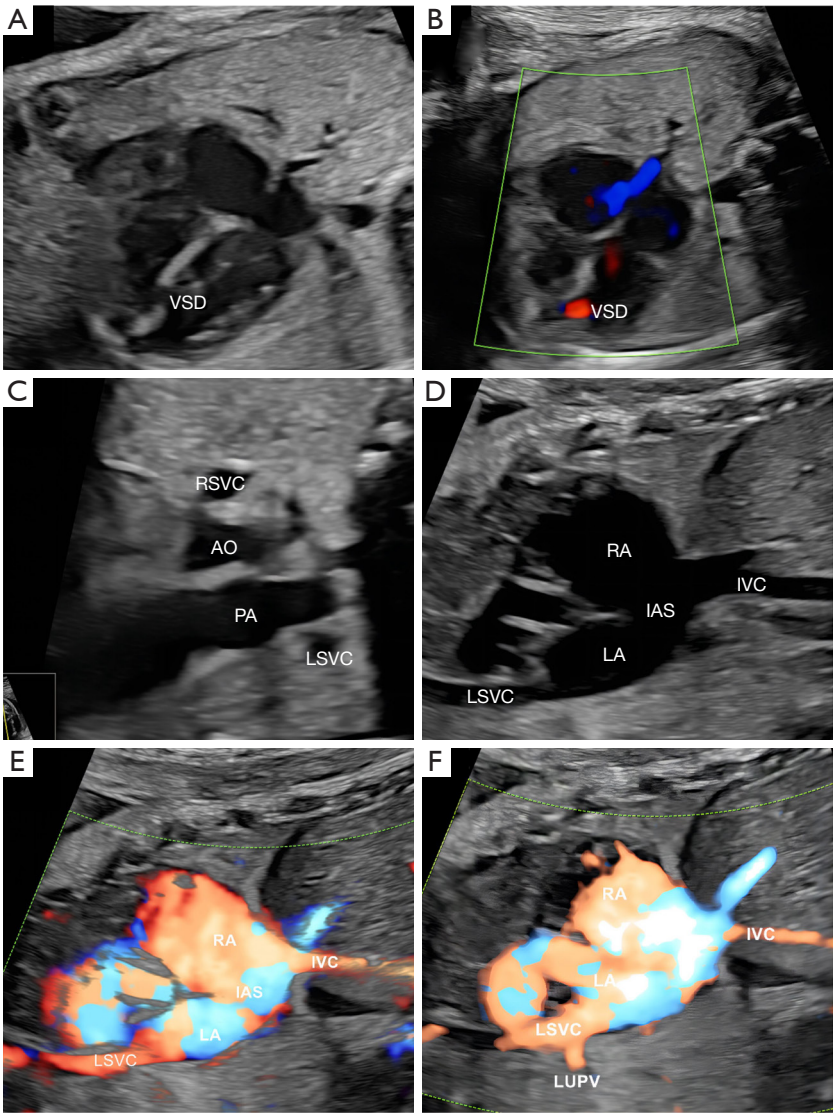

Figure 1 Prenatal echocardiography showed a muscular VSD, LSVC draining into LA and ASD. (A,B) A muscular VSD was visualized using 2D and color Doppler in four-chamber views. (C) To the left of the ductal arch a LSVC was detected in the three-vessel view. (D,E) The left longitudinal view showed that the LSVC drained into the LA; there was also a wide communication between the RA and the LA without a valve of foramen ovale. (F) The opening of the LSVC in LA was in the left superior aspect next to the LUPV. VSD, ventricular septal defect; RSVC, right superior vena cava; AO, aorta; PA, pulmonary artery; LSVC, left superior vena cava; RA, right atrium; IVC, inferior vena cava; IAS, interatrial septum; LA, left atrium; LUPV, left upper pulmonary vein; ASD, atrial septal defect.

left atrial appendage in the parasternal short axial view (*Figure 2C*). Color Doppler showed considerable flow through the LSVC to the LA (*Figure 2D*). A large ASD was also found near the IVC with marked left to right shunt and a maximum diameter of 15.1 mm (*Figure 2E,2F*). The CS was absent. The RA and right ventricle were dilated. A postnatal computed tomography scan confirmed these findings and also showed that the LSVC drained into the LA, with an absent CS (*Figure 3*).

Because of the pulmonary hypertension due to VSD, the patient underwent surgical repair via sternotomy to repair the defects and re-route her LSVC to the RA 9 months after birth. Intraoperative observations confirmed our diagnosis. Following surgery, she completed cardiac rehabilitation. The patient's most recent follow-up showed that she was recovering well. The medical history of patient's parents was

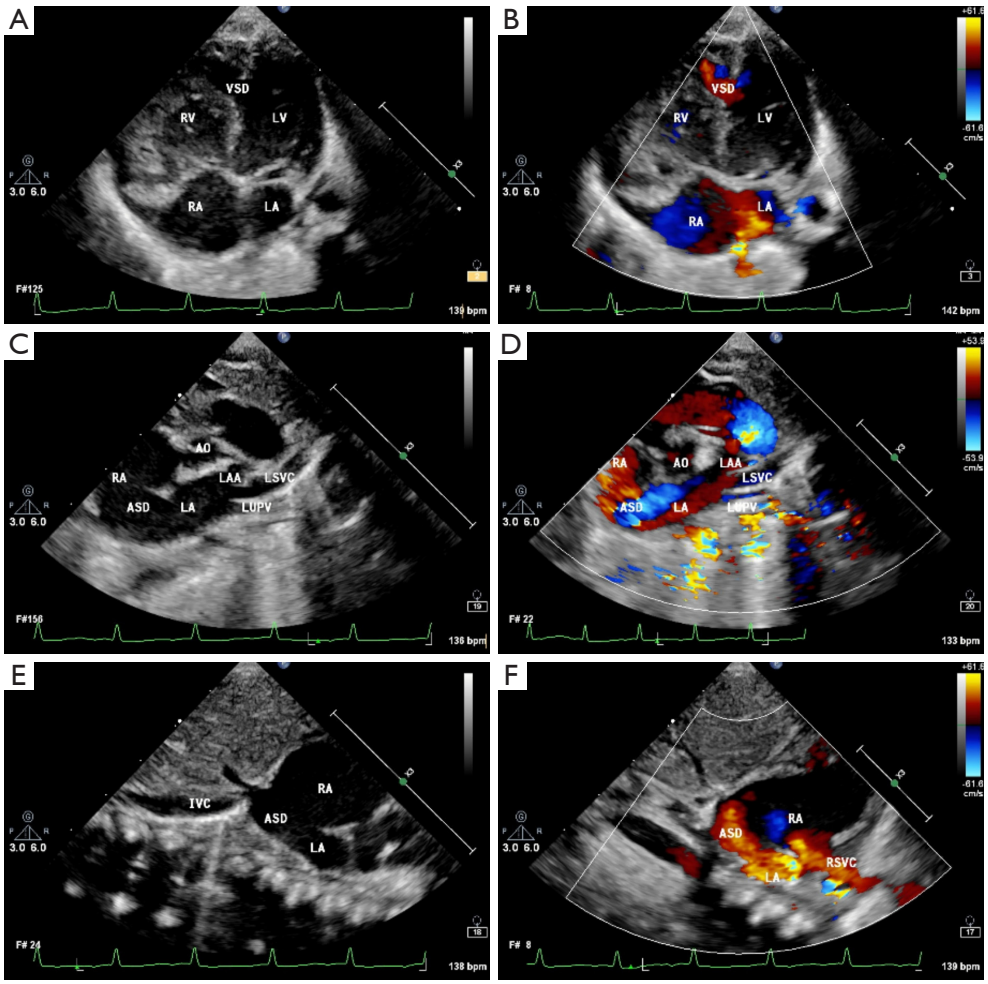

Figure 2 Postnatal echocardiography showed a muscular VSD, LSVC draining into LA and ASD. (A,B) Echocardiography showed muscular VSD in Apical four-chamber view. (C,D) A LSVC returning directly into the upper left corner of LA between the LUPV and LAA in the parasternal short axial view. (E,F) A large ASD was also found near the IVC with marked left to right shunt. VSD, ventricular septal defect; RV, right ventricle; LV, left ventricle; RA, right atrium; LA, left atrium; AO, aorta; LAA, left atrial appendage; LSVC, left superior vena cava; LUPV, left upper pulmonary vein; ASD, atrial septal defect; IVC, inferior vena cava; RSVC, right superior vena cava.

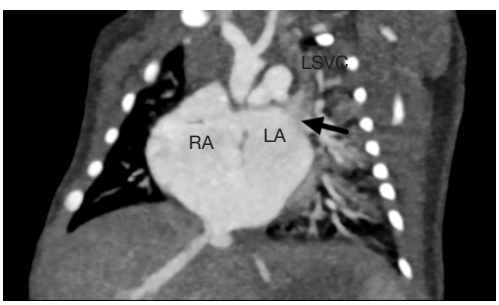

Figure 3 A postnatal computed tomography scan showed the LSVC drained into the LA. Arrow indicates entrance from LSVC to LA. LSVC, left superior vena cava; LA, left atrium; RA, right atrium.

uneventful without congenital abnormalities. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

Discussion

Raghib syndrome is a rare heart malformation that was first reported by Raghib *et al.* in 1965 consisting of three

anomalies (1). In this syndrome, there is termination of a LSVC in the LA, an absence of the CS, and an ASD lying in the posterior lower septum. This syndrome is considered to result from incomplete separation of the sinus venosus from the LA. The incidence of LSVC in the general healthy population is 0.2% to 3% (2). In 90% of the cases, the LSVC drains its deoxygenated blood into the RA by the CS and has no clinical implications. In the remainder of the cases, this vein drains into the LA directly, leading to a right-to-left shunt. Most cases of LSVC draining to the LA have been reported in patients with congenital heart defects (3). In this complex, the left atriovenous fold is underdeveloped, and it may due to be failure of the union of the fold with the septum primum (1). The interatrial communication is considered a sinus venosus septal defect and is accompanied by an absence of the CS.

LSVC can be diagnosed prenatal (2). LSVC is associated with congenital heart disease and other abnormalities. Therefore, prenatal diagnosis of LSVC should be followed by careful screening for other fetal abnormalities. The three-vessel view is important for the prenatal diagnosis of LSVC (4). The three vessels are the arterial duct, aortic arch, and right superior vena cava from left to right normally. In cases of LSVC, the LSVC is prominent on the left side of the ductus arterious (5,6). Additionally, the LSVC returning to the LA and the normal IVC entering the RA could be observed in the left longitudinal view. The use of color and spectral Doppler enables further diagnostic accuracy.

The prenatal diagnosis of ASD is often difficult because of a normal opening of fetal foramen ovale (6). A wide communication between the RA and LA without a valve of foramen ovale may indicate ASD. The absence of atrial septal tissue near the IVC also confirms the location of the defect in the left longitudinal view. The findings in this case emphasize the importance of this view in evaluating such a rare congenital heart disease.

Most patients with Raghib syndrome have few, if any, symptoms. Clinically, features such as increased pulmonary blood flow and skin cyanosis, are often observed in this syndrome. However, serious complications due to LSVC draining into the LA have been reported, including reduced exercise tolerance, syncope, progressive fatigue and severe cyanosis. These patients are prone to thrombotic events and even brain abscesses (7). Additionally, LSVC draining into the LA can cause serious complications during vascular interventional procedures or surgical treatment of cardiac anomalies (2). However, diagnosing Raghib syndrome by clinical symptoms and signs only is difficult (7). In cases of LSVC, a detailed fetal echocardiography is required to confirm the reflux patterns of the LSVC in different views and to exclude other possible concomitant structural malformations. Following a prenatal diagnosis of this syndrome, perinatal management may be optimized. Pregnant women can choose to give birth in a hospital with neonatal care unit; doctors could detect the neonatal hypoxic state at an early stage; accurate diagnosis could be made and the right timing of surgery could be determined.

Raghib syndrome should be suspected in cases of left-to-right interatrial shunt and unexplained hypoxemia or brain abscess (8). Potential treatment strategies include observation or surgery. The principle of treatment timing for this malformation remains unclear. However, surgery is recommended in patients with significant hypoxemia and shunt (9). Sibley *et al.* reported the youngest case of Raghib syndrome in which there was successful surgical correction of an isolated LSVC returning to the LA with a hypoplastic right superior vena cava (10).

The findings in this case show that even prenatal detection of Raghib syndrome may be possible. The prenatal diagnosis of Raghib syndrome may rely on the three-vessel view and the left longitudinal view. A detailed echocardiographic evaluation may be necessary to support this diagnosis. The optimal treatment strategy for Raghib syndrome is surgical repair

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Footnote

Conflicts of Interest: Both authors have completed the ICMJE uniform disclosure form (available at https://qims.amegroups.com/article/view/10.21037/qims-22-465/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical

standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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