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Original Article

Absent right superior caval vein in situs solitus

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Abstract *Introduction:* In up to 0.07% of the general population, the right anterior cardinal vein obliterates and the left remains open, creating an absent right superior caval vein and a persistent left superior caval vein. Absent right superior caval vein is associated with additional congenital heart disease in about half the patients. We wished to study the consequences of absent right superior caval vein as an incidental finding on prenatal ultrasonic malformation screening. *Material and methods:* This is a retrospective case series study of all foetuses diagnosed with absent right superior caval vein at the national referral hospital, Rigshospitalet, Denmark, from 2009 to 2012. *Results:* In total, five cases of absent right superior caval vein were reviewed. No significant associated cardiac, extra-cardiac, or genetic anomalies were found. Postnatal echocardiographies confirmed the diagnosis and there were no postnatal complications. All children were found to have healthy hearts at follow-up. *Conclusions:* In all cases, the findings proved to be a benign condition with no clinical manifestations or complications. Although isolated absent right superior caval vein does not seem to affect the outcome, associated anomalies may be serious. Absent right superior caval vein should, therefore, prompt a search for additional malformations. Furthermore, the diagnosis of an isolated absent right superior caval vein is important, because knowledge of the anomaly can prevent future problems when invasive procedures are necessary.

Keywords: Congenital heart disease; absent right superior caval vein; foetal echocardiography

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DURING EMBRYONIC LIFE, THE VENOUS BLOOD enters the heart through the cardinal veins and sinus venosus. Owing to drainage of venous blood to the right side through the innominate vein, the right sinus horn and anterior cardinal vein ultimately become the superior caval vein, whereas the left sinus horn and anterior cardinal vein are diminished and form the coronary sinus (Fig 1).

A persistent left superior caval vein is the most common thoracic venous anomaly[1–3] and is found when the left anterior cardinal vein fails to close. In 2.4% of foetuses with persistent left superior caval vein,[4] the right anterior cardinal vein obliterates (Fig 1). The left superior caval vein is then fed through retrograde flow in the brachiocephalic vein and empties into the coronary sinus. This in turn drains into the right atrium, except in 1–10% of cases where the coronary sinus remains unroofed and communicates with the left atrium,[2,5–7] which in turn leads to arterial desaturation due to the shunting of blood from right to left.[2,6] Absent right and persistent left superior caval vein are found in 0.05–0.07% of the general population and in 0.007–0.14% of hearts with congenital cardiovascular malformations.[5,7–10]

Foetal echocardiography can in the three-vessel view visualise the absent right superior caval vein with persistent left superior caval vein. In the normal three-vessel view, the pulmonary artery, the aortic arch, and the right superior caval vein form a line from left to right. In cases with absent right and persistent left superior caval vein, the three-vessel view also shows three vessels in a line, but the left

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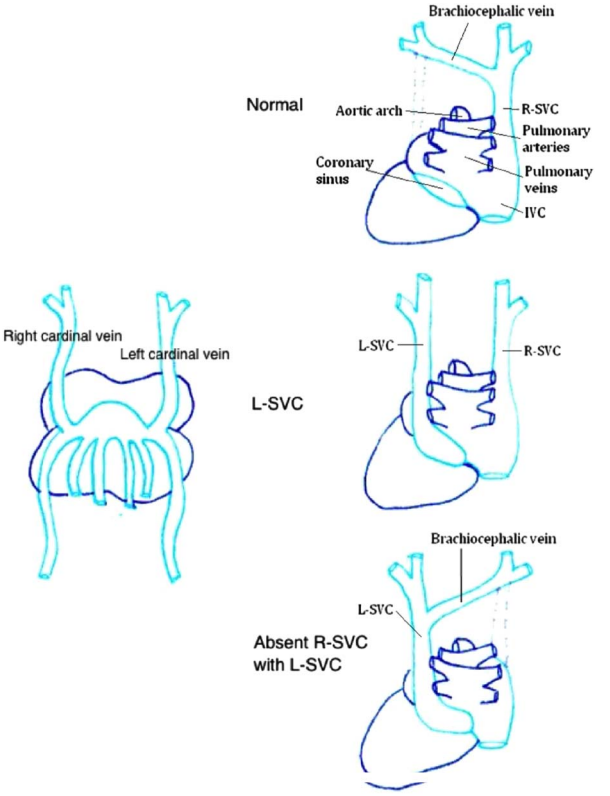

Figure 1.
Development of the superior caval vein (SVC) in the structurally normal heart, persistent left SVC, and absent right with persistent left SVC.

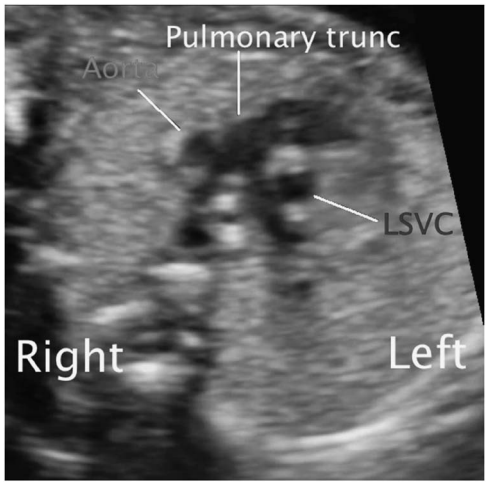

Figure 2.
Three-vessel view of the absent right superior caval vein in situs solitus. This view can give the false impression of transposition of the great arteries.

superior caval vein is to the left and the aortic arch is to the right (Fig 2). Another marker is the dilated coronary sinus.[2,5,10–11] In the four-chamber view, this anomaly can be mistaken for an

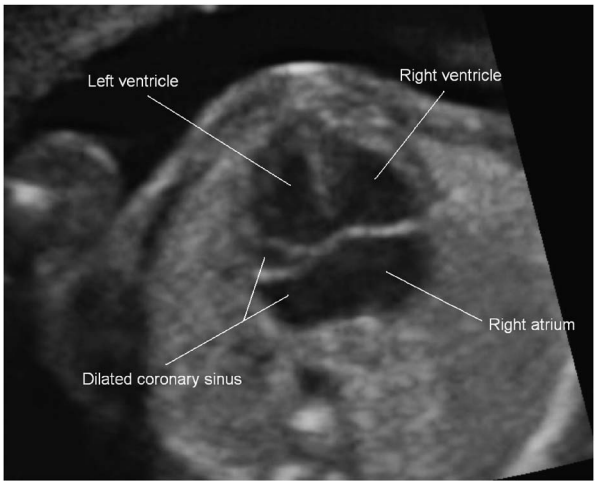

Figure 3.
Four-chamber view at the level of the large coronary sinus that gives the appearance of atrioventricular septal defects. The AV-valves were structurally normal and there was no ventricular septal defect detected after delivery.

atrioventricular septal defect or an inlet ventricular septal defect,[12] (Fig 3).

Absent right superior caval vein is associated with additional congenital heart disease in about half the patients.[9] It is commonly associated with cardiac situs anomalies[7,11] and seems to be associated with atrial septal defects, atrioventricular septal defect and tetralogy of Fallot, as well as arrhythmias.[9] Furthermore, persistent left superior caval vein is associated with a relative obstruction of the left atrioventricular inflow caused by the dilated coronary sinus,[2,9,12] subsequently resulting in aortic stenosis and coarctation of the aorta.

We wished to study the consequences of absent right superior caval vein in situs solitus as an incidental finding on prenatal ultrasonic malformation screening at the national referral hospital of Denmark. Our aim was to familiarise obstetric sonographers and paediatric cardiologists with the condition as well as its consequences and pitfalls. To our knowledge, although the condition in adults is well-described in the literature, the consequences of diagnosing it prenatally have not been adequately addressed.

Material and methods

This is a retrospective case series study of all foetuses diagnosed with absent right superior caval vein at the national referral hospital, Rigshospitalet, in Denmark from 2009 to 2012. No patients were lost to follow-up. Rigshospitalet conducts standard prenatal ultrasound screening as well as specialised foetal echocardiography. If standard screening at regional hospitals raises suspicion of cardiac malformations, the patient is referred to Rigshospitalet where foetal

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Table 1. Associated cardiac anomalies and outcome in five cases of absent right and persistent left superior caval vein.

Case no.	Sex	Prenatal echocardiography	Postnatal echocardiography	ECG	Age at follow-up (months)	Findings at follow-up
1	F	HAA, VSD, dCS	–	VES*	3	Healthy, no ass. cardiac anomalies; Still's murmur, VES*
2	M	HMV, HAV, HAA, VD, dCS	VSD, VD, PH, PDA, dCS	RVH	3	Healthy, no ass. cardiac anomalies
3	M	HMV, HAV, HAA, VD, dCS	VD, ASD, PDA with L–R shunt, dCS	N/A	1	Healthy, no ass. cardiac anomalies
4	M	HAA, VD, CoA, TI	PFO, VSD	N/A	3	Healthy, no ass. cardiac anomalies
5	F	dCS	dilated LA, VD, PFO and PDA with L–R shunt, dCS	N/A	1	Healthy, no ass. cardiac anomalies

ass. = associated; CoA = coarctation of the aorta; dCS = dilated coronary sinus; HAA = hypoplastic aortic arch; HAV = hypoplastic aortic valve; HMV = hypoplastic mitral valve; LA = left atrium; L–R = left to right; PH = pulmonary hypertension; PDA = persistant ductus arteriousus; PFO = persistant foramen ovale; RVH = right ventricular hypertrophy; TI = triscupid insufficiency; VD = ventricular disproportion; VES = ventricular extrasystoles; and VSD = ventricular septal defect

*Re-referred for evalutation at 3 years of age due to irregular heart beat and heart murmur

echocardiography is carried out by trained foetal cardiologists.

All patients diagnosed with absent right superior caval vein during this period were included. The mothers' obstetric records as well as prenatal ultrasounds were reviewed, and prenatal findings along with obstetric complications were described. Invasive procedures and genetic investigations, if any, were reported.

The newborns' medical records were reviewed and the method of delivery, gestational age, and complications during delivery were recorded. The neonatal period was described and any symptoms or objective findings were reported. Postnatal ultrasounds were assessed and compared with the prenatal ultrasounds.

Follow-up was carried out at 1–3 months of age. The medical records were reviewed for any contact with the hospital during the following 1–4 years.

Ethical protocol was followed.

Results

Over a period of 4 years, five foetuses were diagnosed with absent right superior caval vein. In three cases, the anomaly was an incidental finding during a standard prenatal ultrasonic screening for malformations at gestational age 19–22 weeks. In cases 2 and 3, the women were referred to foetal echocardiography as the standard screening had raised suspicions of a cardiac anomaly. The atrial arrangement was situs solitus in all five foetuses, all atrioventricular and ventriculoarterial connections were concordant and no extra-cardiac anomalies were detected.

In this study, we describe the prenatal and postnatal findings in the individual cases. Prenatal and postnatal cardiac anomalies in each patient are described in Table 1.

Case 1

During a standard prenatal ultrasound at gestational age 20 + 3, the foetus was found to have an absent right and persistent left superior caval vein. Foetal echocardiography performed the same day confirmed the diagnosis.

The flow in the brachiocephalic vein was right to left. The left caval vein emptied into a dilated coronary sinus, but there was no obstruction of inflow to the left ventricle.

The left ventricle and mitral valve were of normal size, but the aortic arch was assessed to be slightly smaller than normal.

Furthermore, the foetus was suspected of having minor muscular ventricular septal defects.

Follow-up foetal echocardiography every 3 weeks due to oligohydramnios for the remainder of the pregnancy showed no change in the condition, and as heart function was found to be normal no further measures were taken.

Amniocentesis was carried out according to the local protocol and the genetic analysis was normal.

The girl was born at full term and was not admitted to the Neonatal Intensive Care Unit for observation after delivery. Postnatal echocardiography at 2 months confirmed the diagnosis of absent right superior caval vein, but no ventricular septal defect could be identified. Ventricles, valves, and aortic arch were of normal size, but the coronary sinus was dilated.

Final follow-up echocardiography was carried out a year later and no development in the condition was found.

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During a standard child examination at age 3, an irregular heart rate and heart murmur were found. After tests had been carried out, these were found to be extrasystoles and a Still's murmur, and no further actions were taken.

Case 2

The diagnosis was made at gestational age 22+5. Although the brachiocephalic vein could not be visualised, it was assumed that the flow would have to be retrograde. No other anomaly was found; however, during follow-up foetal echocardiography at 28 and 34 weeks of gestation, the mitral and aortic valves were found to be considerably smaller than the tricuspid valve and the pulmonic valve (z-scores, respectively, −4.1 and −6.4 versus −0.9 and 0.7). The aortic arch was deemed hypoplastic (z-score unavailable). The left ventricle was evaluated to be smaller than the right ventricle, but its function was normal.

No genetic analysis was carried out.

The boy was born at full term and received full Apgar Scores at 1 and 5 minutes. Postnatal echocardiography after 24 hours revealed absent right and persistent left superior caval vein, a normal aortic arch, an insignificant persistent arterial duct, and a small muscular ventricular septal defect. The right ventricle was slightly dilated with a tricuspid insufficiency gradient of 50 mmHg. Electrocardiogram showed right ventricular hypertrophy. The boy, however, had no respiratory problems, saturated 100% without supplementary oxygen, and was discharged from the Neonatal Intensive Care Unit. Additional echocardiographies showed full regression of pulmonary hypertension and ventricular septal defect at 3 months of age.

Case 3

At gestational age 21+1, the foetus was found to have absent right and persistent left superior caval vein. Flow from right to left was seen in the brachiocephalic vein, draining into the left superior caval vein, which in turn emptied into a dilated coronary sinus. There was no obstruction of the left atrioventricular inflow and no ventricular disproportion or dysfunction was observed.

Follow-up foetal echocardiography at 30 weeks of gestation revealed a subjectively slightly hypoplastic mitral valve, aortic valve, and aortic arch as well as a small left ventricle with normal function.

The foetus also presented with polyhydramnios and macrosomia, and flow measurements and weight scans were carried out every 2 weeks for the remainder of the pregnancy. No gestational diabetes, foetal hydrops, or incompensation were found and therapeutic amniocentesis was carried out once. At this time, genetic analysis was carried out according to local protocol, and the karyotype was found to be normal.

The boy was delivered by caesarean section at gestational age 38+1 because of macrosomia. His Apgar score was 9 at 1 minute and 10 at 5 minutes. Postnatal echocardiography performed the same day confirmed the diagnosis and showed a left ventricle that was slightly smaller than the right, but with normal function. Follow-up echocardiography 4 days later revealed a small atrial septal defect with a left-to-right shunt. He was discharged from the Neonatal Intensive Care Unit after 4 days of observation. Final echocardiography after 2 months did not show an atrial septal defect or a small left ventricle.

Case 4

The diagnosis was suspected during a routine scan at gestational age 20+0, and was confirmed by foetal echocardiography the next day. Retrograde flow was found in the brachiocephalic vein, which emptied into the left caval vein. The caval vein drained into the coronary sinus, which was not found to be dilated.

Discrete hypoplasia of the aortic arch was noted, as the diameter was 50% of that of the pulmonary trunck. The left ventricle was of normal size and function.

At week 26 of gestation, there was some ventricular disproportion, the left ventricle being subjectively smaller than the right, but its function was still normal. At 31 weeks of gestation, the foetus had signs of coarctation of the aorta, and at 39 weeks slight tricuspid insufficiency.

Genetic analysis by amniocentesis offered according to local protocol was normal.

The boy was born at gestational age 41+3, and received normal Apgar scores of 9 and 10 at 1 and 5 minutes, respectively. He received treatment with continuous positive airway pressure for 5 minutes due to grunting. Pulse oximetry was normal.

At the Neonatal Intensive Care Unit, a systolic heart murmur was found. Echocardiography 3 days after birth revealed an insignificant persistent foramen ovale and a possible ventricular septal defect, but no signs of coarctation of the aorta. Neither foramen ovale nor a ventricular septal defect were found during the follow-up echocardiography at 2 months of age, and the child did not have a heart murmur.

Case 5

Case 5 was a twin pregnancy. Twin A was, at gestational age 19+0, found to have absent right and persistent left superior caval vein, which was fed through the braciocephalic vein and emptied into the

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coronary sinus. Twin B had no heart anomalies. Follow-up foetal echocardiography was carried out at 21 and 28 weeks of gestation, and the condition was stationary with no hypoplasia of the aortic arch, left ventricle, or valves.

Flow measurements and weight scans were carried out every week for the remainder of the pregnancy.

The risk for Down's syndrome was greater than 1:300 due to an enlarged nuchal fold, wherefore chorionic villus biopsy was secured at 12 weeks of gestation and was found to be normal.

The girl was delivered by acute caesarean section at gestational age 34+3, as the mother developed severe preeclampsia. Apgar score was 8 at 1 minute and 10 at 5 minutes. Shortly after birth, she developed respiratory distress. She received treatment with continuous positive airway pressure and supplementary oxygen and was admitted to the Neonatal Intensive Care Unit. After 6 days, she no longer needed continuous positive airway pressure.

Postnatal echocardiography after 24 hours confirmed the diagnosis of absent right superior caval vein and showed a slightly dilated left atrium with a persistent foramen ovale with a left-to-right shunt. There was also a persistent arterial duct with bi-directional flow.

After 10 days, follow-up echocardiography showed no persistent arterial duct or foramen ovale and the girl was discharged after 4 weeks.

Discussion

We have presented five cases of absent right with persistent left superior caval vein in situs solitus diagnosed prenatally. To our knowledge, this is the largest collection of absent right superior caval vein diagnosed in foetuses. In all cases, the finding proved to be a benign condition with no clinical manifestations or complications. This is in keeping with previous reports of isolated absent right superior caval vein found as an incidental finding at autopsy, during pre-surgical imaging, or during surgical procedures in asymptomatic individuals.[5,10,13]

With the offer of prenatal screening for malformations, more cases of absent right superior caval vein in situs solitus can be diagnosed prenatally, raising questions about the prognosis and implications.

During standard prenatal ultrasonic scan for malformations, the heart is systematically reviewed in the screening for congenital heart defects. This always includes the four-chamber view, but not always the three-vessel view, depending on the experience of the examiner. As mentioned, a dilated coronary sinus can be misinterpreted as an atrioventricular septal defect, and in severe cases it may be misconceived as mitral atresia, as the mitral valve is located anteriorly and may not necessarily be seen in the four-chamber view.[12] Furthermore, a dilated coronary sinus may also be caused by increased pressure in the right atrium, abnormal pulmonary or systemic venous return, or a fistulous connection with the coronary arteries. The finding of a dilated coronary sinus should, therefore, always lead to the evaluation of the three-vessel view.[12,14]

Previous studies have reported associated cardiac anomalies in 46–100% of the cases.[1,8,9] Usually, the associated defect is depicted first, but the absence of the right superior caval vein should prompt a search for additional cardiac malformations, as they will determine the outcome.[12] The direction of blood flow is of great importance, and should the left superior caval vein drain directly into the left atrium surgery is needed regardless of the associated anomalies.[13]

None of our five patients presented with unroofed coronary sinus or anomalies associated with reduced flow through the left ventricle, such as coarctation of the aorta, aortic stenosis, or hypoplasia of the left ventricle; four patients were suspected of small muscular ventricular septal defects prenatally or postnatally, but follow-up echocardiography found the ventricular septum to be intact. Only one patient had pulmonary hypertension, but full regression was found at 3 months of age. Arrhythmias are associated with absent right superior caval vein, but electrocardiograms were only available in two cases – one showed right hypertrophy due to pulmonary hypertension and the other was diagnosed with extrasystoles at the age of 3 years.

Few studies could be found where the association between absent right superior caval vein and extracardiac malformations was investigated, which Barrea et al call to attention.[12] In their study of 27 patients with anomalous systemic venous return, 14 (52%) – including one patient with absent right superior caval vein – had associated extra-cardiac anomalies. Bearing this in mind, although none of our patients had any extra-cardiac malformations, vigilant investigation should be undertaken in these patients.

No genetic anomaly was detected in our case series. This is in keeping with the findings of Barrea et al, who did not find any abnormal karyotype in four patients with absent right superior caval vein;[12] however, they did find five patients (26%) with persistent left superior caval vein with chromosomal anomalies, which corresponds to the 23% reported by Postema et al.[2]

Invasive procedures such as central catheters, pacemaker implantation, and cardiac surgery can be challenging if the right superior caval vein is absent.[2,13,15] Catheter insertion through the left or right subclavian vein will enter the right atrium unexpectedly through the coronary sinus, necessitating

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a longer lead, which may not be available at the time of the procedure.[15] Positioning the lead in the right atrial appendage is challenging, as the opening into the right atrium from the coronary sinus is very close to the right ventricular inlet, and the procedure requires careful manipulation.[16] Furthermore, when the chosen approach of the procedure is impossible due to the anomaly, the necessary adaptation of the procedure may increase the risk of arrhythmias[11,17] or other complications such as puncture of the atrium[16] or the liver capsule.[17] During open-heart surgery, it is important to bear in mind the changed anatomy when selecting perfusion techniques to ensure adequate venous drainage and myocardial protection.[6,7,11,13]

With the increased use of central catheters, pacemaker implantation, and cardiac surgery, knowledge of this anomaly is important to prevent future complications and morbidity.

Conclusion

Isolated absent right superior caval vein is frequently a benign condition, but when it is diagnosed prenatally it is important to carry out investigations to detect any associated anomalies, this being intra-cardiac, extra-cardiac, or chromosomal, as this is crucial to guide parental counselling and management appropriately. Furthermore, diagnosis of isolated absent right superior caval vein is important, because knowledge of the anomaly can prevent future problems when invasive procedures are necessary.

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Conflicts of Interest

None.

Ethical Standards

This research followed national guidelines for ethical standards.

References

1. Choi JY, Anderson RH, Macartney FJ. Absent right superior caval vein (caval vein) with normal atrial arrangement. Br Heart J 1987; 57: 474–478.
2. Postema PG, Rammeloo LA, van Litsenburg R, Rothuis EG, Hruda J. Left superior vena cava in pediatric cardiology associated with extra-cardiac anomalies. Int J Cardiol 2008; 123: 302–306.
3. Yuce M, Kizilkan N, Kus E, Davutoglu V, Sari I. Giant coronary sinus and absent right superior caval vein. Vasa 2011; 40: 65–67.
4. Berg C, Knüppel M, Geipel A, et al. Prenatal diagnosis of persistent left superior caval vein and its associated congenital anomalies. Ultrasound Obstet Gynecol 2006; 27: 274–280.
5. Sheikh AS, Mazhar S. Persistent left superior vena cava with absent right superior vena cava: review of the literature and clinical implications. Echocardiography 2014; 31: 674–679.
6. Neema PK, Manikandan S, Rathod RC. Absent right superior vena cava and persistent left superior vena cava: the perioperative implications. Anesth Analg 2007; 105: 40–42.
7. Buirski G, Jordan SC, Joffe HS, Wilde P. Superior vena caval abnormalities: their occurrence rate, associated cardiac abnormalities and angiographic classification in a paediatric population with congenital heart disease. Clin Radiol 1986; 37: 131–138.
8. Lenox CC, Zuberbuhler JR, Park SC, et al. Absent right superior caval vein with persistent left superior caval vein: implications and management. Am J Cardiol 1980; 45: 117–122.
9. Bartram U, Van Praagh S, Levine JC, Hines M, Bensky AS, Van Praagh R. Absent right superior caval vein in visceroatrial situs solitus. Am J Cardiol 1997; 80: 175–183.
10. Waikar HD, Lahie YK, De Zoysa L, Chand P, Kamalanesan RP. Systemic venous anomalies: absent right superior caval vein with persistent left superior caval vein. J Cardiothorac Vasc Anesth 2004; 18: 332–335.
11. Gologorsky E, Macedo FI, Carvalho EM, Govindaswamy R, Salerno TA. Unexpected persistent left superior vena cava and absent right superior vena cava in situs solitus patient. J Card Surg 2010; 25: 42–45.
12. Barrea C, Ovaert C, Moniotte S, Biard JM, Steenhaut P, Bernard P. Prenatal diagnosis of abnormal cardinal systemic venous return without other heart defects: a case series. Prenat Diagn 2011; 31: 380–388.
13. Luo ZQ, Liu KY, Han Z, Zhou C, Liu FL, Zhou XD. Surgical management of persistent left superior vena cava associated with an absent right superior vena cava. J Card Surg 2012; 27: 117–118.
14. Uçar O, Paşaoğlu L, Çiçekçioğlu H, Vural M, Kocaoğlu I, Aydoğdu S. Persistent left superior vena cava with absent right superior vena cava: a case report and review of the literature. Cardiovasc J Afr 2010; 21: 164–166.
15. Alemzadeh-Ansari MJ, Shafiee A, Yaminisharif A. Implantation of transvenous permanent pacemaker in a patient with persistent left superior vena cava and absent right superior vena cava. Int Cardiovasc Res J 2013; 7: 152–154.
16. Rizkallah J, Burgess J, Kuriachan V. Absent right and persistent left superior vena cava: troubleshooting during a challenging pacemaker implant: a case report. BMC Res Notes 2014; 7: 462.
17. Chovanec V, Krajina A, Hůlek P, Mest'an M, Renc O. TIPS creation in patients with persistent left superior vena cava. Acta Medica (Hradec Kralove) 2010; 53: 35–38.