v. 4, n. 19, 2024

International Journal of Health Science

RIGHT SUPERIOR VENA CAVA AGENESIS AND PERSISTENT LEFT SUPERIOR VENA CAVA: LITERATURE REVIEW AND CASE REPORT

Andrés Sebastián Viteri Hinojosa

All content in this magazine is licensed under a Creative Commons Attribution License. Attribution-Non-Commercial-Non-Derivatives 4.0 International (CC BY-NC-ND 4.0).

Abstract: Right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) are uncommon anomalies arising from faulty embryonic development. This review discusses epidemiology, clinical presentations, diagnostic methods, treatment options, follow-up strategies, and emerging perspectives. RSVC agenesis stems from an underdeveloped right anterior cardinal vein, while PLSVC signifies the persistence of the left counterpart, creating an additional drainage pathway to the right atrium. Their clinical spectrum ranges from asymptomatic discoveries to potential complications like arrhythmias, thrombosis, or catheter-related issues. Diagnosis relies on imaging tools like echocardiography, CT, MRI, or venography. Treatment choices, influenced by individual presentations and complications, include conservative management, anticoagulation, and surgical correction. Long-term follow-up remains crucial for monitoring potential complications and tailoring management. New vistas on the horizon involve advancements in imaging, genetic studies, and minimally invasive treatments. Case studies like the one presented also contribute valuable insights, expanding our understanding of these rare venous anomalies. In conclusion, RSVC agenesis and PLSVC pose unique diagnostic and management challenges due to their rarity and variable presentations. Continued research and collaboration are essential to reveal their pathophysiology, improve diagnostic accuracy, and optimize treatment outcomes for individuals with these congenital venous anomalies.

INTRODUCTION

Right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) represent uncommon congenital anomalies of the thoracic venous system, affecting an estimated 0.3% to 0.5% of the general population [3]. Despite their modest prevalence, these anatomical variations pose unique challenges in diagnosis and management due to their rarity and diverse clinical presentations [4]. This review delves into the current understanding of RSVC agenesis and PLSVC, providing an overview of their epidemiology, clinical manifestations, diagnostic modalities, treatment strategies, follow-up approaches, and emerging perspectives.

Both RSVC agenesis and PLSVC arise from disruptions in embryologic development of the cardinal veins, the precursors to the superior vena cavae [3]. During fetal development, three pairs of cardinal veins (anterior, common, and posterior) drain the upper body into the heart. Typically, the right anterior cardinal vein regresses, while the left anterior cardinal vein incorporates into the coronary sinus and left brachiocephalic vein, ultimately forming the left superior vena cava [5]. However, in RSVC agenesis, the right anterior cardinal vein fails to develop properly, leading to the absence or hypoplasia of the right superior vena cava [2]. Conversely, PLSVC results from the persistence of the left anterior cardinal vein, creating an additional venous pathway draining directly into the right atrium or coronary sinus [7].

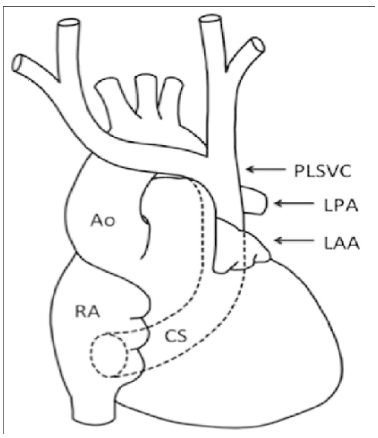
EPIDEMIOLOGY

Right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) are rare congenital anomalies of the venous system, with reported incidences varying across different populations. The prevalence of RSVC agenesis has been estimated to range from 0.3% to 0.5% in the general population, while the prevalence of PLSVC is reported to be approximately 0.3% to 0.5% as well (1,2).

These anomalies are often detected incidentally during diagnostic imaging studies or medical procedures. For instance,

a study by Aydin et al. reported the incidental finding of absent RSVC and persistent LSVS in 0.046% of cases during transthoracic echocardiography (4). Similarly, Goyal et al. described encountering this rare anatomical variant during central venous line placement in clinical practice (3).

While RSVC agenesis and PLSVC can occur independently, there are also cases where they coexist. Double superior vena cava (DSVC) with absent RSVC and persistent LSVC is a rare variation, as highlighted in a case report by Kumar et al. (1). The coexistence of these anomalies presents unique challenges in diagnosis and management, requiring a thorough understanding of their anatomical variations and potential clinical implications.



Interestingly, these anomalies have been reported across different age groups, from infants to adults. Elgendy et al. presented a case of absent RSVC with persistent LSVC diagnosed in a 27-year-old patient through contrast-enhanced computed tomography (5). Similarly, Jindal et al. reported an unusual anatomical variation of the superior vena cava system in a 45-year-old patient diagnosed on computed tomography angiography (6).

The association of RSVC agenesis and PLSVC with other congenital heart defects or syndromes has also been documented. For example, van der Horst et al. reported a case of isolated absence of the right superior vena cava with persistent left superior vena cava in a patient with transposition of the great arteries (15). These associations underscore the importance of comprehensive cardiac evaluation and genetic screening in individuals diagnosed with these venous anomalies.

Overall, while RSVC agenesis and PLSVC are rare anatomical variants, their detection is increasing due to advancements in diagnostic imaging modalities and greater awareness among healthcare providers. Further epidemiological studies are warranted to better understand the prevalence, associated risk factors, and clinical implications of these congenital venous anomalies across different populations.

CLINICAL MANIFESTATIONS

The clinical manifestations of right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) vary widely, ranging from asymptomatic incidental findings to potentially life-threatening complications. Many individuals with these venous anomalies remain asymptomatic throughout their lives, with the condition often discovered incidentally during diagnostic imaging or medical procedures (4, 7).

However, some patients may present with symptoms related to associated cardiovascular abnormalities or complications. Common symptoms include dyspnea, chest pain, palpitations, and syncope. These symptoms may result from underlying cardiac conditions such as atrial septal defects, ventricular septal defects, or arrhythmias (11). Additionally, thrombosis or catheter-related issues can lead to symptoms such as arm swelling, pain, or discomfort at the site of central venous catheter insertion (3).

Complications associated with RSVC agenesis and PLSVC include thrombosis, embolism, and arrhythmias. Thrombosis can occur within the anomalous venous pathway

or in the presence of central venous catheters, particularly in patients undergoing long-term intravenous therapy or hemodialysis (3, 7). Embolism, including pulmonary embolism, is a potentially life-threatening complication that may arise from thrombus formation within the venous system (9).

Arrhythmias such as atrial fibrillation or atrial flutter are also commonly reported in patients with RSVC agenesis and PLSVC. These arrhythmias may be attributed to abnormal cardiac conduction pathways or accessory pathways associated with congenital heart defects (1, 6). Additionally, the presence of an anomalous venous pathway may increase the risk of arrhythmias during invasive cardiac procedures or catheter ablation therapies (8).

DIAGNOSIS

The diagnosis of RSVC agenesis and PLSVC typically involves a combination of clinical evaluation and diagnostic imaging studies. Transthoracic echocardiography (TTE) is often used as an initial screening tool to assess cardiac anatomy and function. In cases where venous anomalies are suspected, TTE may reveal abnormal venous connections or flow patterns suggestive of RSVC agenesis or PLSVC (4).

Further imaging studies such as computed tomography (CT) angiography, magnetic resonance imaging (MRI), or venography may be required to confirm the diagnosis and delineate the anatomical details of the venous system. Contrast-enhanced CT or MRI allows for visualization of the venous anatomy and can accurately identify the presence of RSVC agenesis, PLSVC, or other associated anomalies (5, 12).

Invasive procedures such as cardiac catheterization may be indicated in select cases to assess hemodynamic parameters or perform interventions such as closure of associated cardiac defects. Preprocedural imaging of the superior vena cava anatomy is essential to guide catheterization procedures and minimize the risk of complications such as vascular injury or arrhythmias (14).

Genetic testing may also be considered in patients with RSVC agenesis and PLSVC, particularly if there is suspicion of an underlying genetic syndrome or association with other congenital anomalies. However, the genetic basis of these venous anomalies remains poorly understood, and further research is needed to elucidate the underlying pathogenesis and genetic risk factors (13).

TREATMENT

The management of right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) depends on the clinical presentation, associated complications, and patient-specific factors. Treatment strategies may include conservative measures, pharmacological interventions, or surgical procedures to prevent complications and improve patient outcomes.

In asymptomatic individuals with incidental findings of RSVC agenesis or PLSVC, conservative management may be sufficient, focusing on regular monitoring and surveillance to detect potential complications such as thrombosis or arrhythmias. This approach is supported by the low incidence of symptomatic complications in patients with these venous anomalies (2, 7).

Pharmacological interventions such as anticoagulation therapy may be indicated in patients at high risk of thrombosis, particularly those with central venous catheters or other predisposing factors. Anticoagulants such as heparin or warfarin may be prescribed to prevent thrombus formation and reduce the risk of embolic events (3, 9).

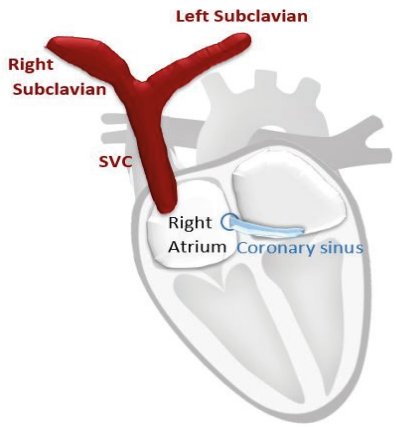
In cases where symptomatic complications arise, such as thrombosis or arrhythmias, more aggressive treatment strategies may

be necessary. Thrombolytic therapy or surgical thrombectomy may be considered for the management of venous thrombosis, particularly in patients with extensive or life-threatening thrombus burden (6, 11).

Arrhythmias associated with RSVC agenesis and PLSVC may require pharmacological management with antiarrhythmic medications or invasive procedures such as catheter ablation. Cardiac electrophysiology studies may be performed to identify abnormal conduction pathways or accessory pathways contributing to arrhythmia genesis (8).

Surgical intervention may be indicated in select cases to correct associated cardiac defects or alleviate symptomatic complications. For example, closure of atrial or ventricular septal defects may be performed to prevent recurrent embolic events or reduce the risk of arrhythmias (1, 15).

Minimally invasive procedures such as transvascular closure of persistent left superior vena cava fenestrations using devices such as Amplatzer duct occluders have also been described in the literature. These techniques offer a less invasive alternative to surgical correction and may be particularly suitable for patients with isolated persistent left superior vena cava fenestrations (8).



FOLLOW UP

Long-term follow-up is essential for patients with RSVC agenesis and PLSVC to monitor for potential complications, assess treatment efficacy, and optimize management strategies. Follow-up care may involve regular clinical evaluations, imaging studies, and cardiac monitoring to detect and address any emerging issues.

Patients with central venous catheters or other predisposing factors for thrombosis should undergo periodic surveillance imaging to screen for venous thrombosis and assess catheter patency. Duplex ultrasonography or venography may be used to evaluate venous blood flow and detect thrombus formation within the anomalous venous pathway (4, 14).

Cardiac monitoring may be indicated for patients with arrhythmias or conduction abnormalities associated with RSVC agenesis and PLSVC. Holter monitoring or event recording may be used to assess cardiac rhythm and identify any recurrent arrhythmias requiring intervention (8).

Regular follow-up visits with a cardiologist or cardiovascular specialist are recommended to ensure continuity of care and address any concerns or questions regarding the patient's condition. Patient education regarding signs and symptoms of potential complications and lifestyle modifications to reduce cardiovascular risk factors is an integral component of long-term management (13).

NEW PERSPECTIVES

Advancements in imaging techniques, genetic studies, and minimally invasive treatment modalities have contributed to new perspectives in the understanding and management of right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC). These developments offer opportunities for improved diagnosis, personalized treatment approaches, and

enhanced patient outcomes.

Imaging modalities such as computed tomography (CT) angiography, magnetic resonance imaging (MRI), and echocardiography play a crucial role in diagnosing and characterizing venous anomalies. These techniques provide detailed anatomical information, allowing for accurate identification of RSVC agenesis, PLSVC, and associated cardiovascular abnormalities (4, 5).

Genetic studies have shed light on the underlying pathogenesis of RSVC agenesis and PLSVC, although the genetic basis of these anomalies remains incompletely understood. Identification of genetic risk factors may help elucidate the etiology of venous anomalies and facilitate early detection and intervention in at-risk individuals (13).

Minimally invasive treatment modalities, such as transvascular closure of persistent left superior vena cava fenestrations using devices like Amplatzer duct occluders, offer a less invasive alternative to traditional surgical approaches. These techniques effectively close anomalous venous pathways while minimizing procedural risks and recovery time (8).

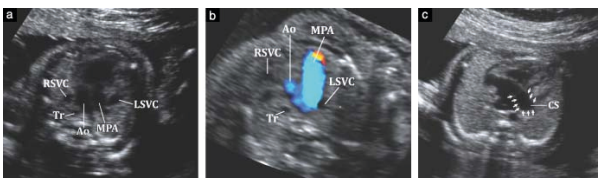
Collaborative research efforts and multidisciplinary approaches are essential to further advance our understanding of RSVC agenesis and PLSVC and improve patient care. Continued research into the genetic basis, pathophysiology, and natural history of venous anomalies will enhance our ability to effectively diagnose and manage these conditions.

PRESENT THE CASE

A 4-year-old patient presented to the community clinic for routine follow-up, with a history of dyspnea on exertion graded as New York Heart Association (NYHA) class III. The patient's medical history included evaluation for an innocent murmur without follow-up. Physical examination revealed a holosystolic murmur (Grade II) predominantly heard at the tricuspid focus.

Further evaluation with a 12-lead electrocardiogram showed evidence of right ventricular overload and left axis deviation, considered normal for the patient's age. Given the clinical presentation, an echocardiogram was performed, revealing situs solitus, levocardia, and levoapex with no structural defects.

Notably, the echocardiogram confirmed agenesis of the right superior vena cava and a persistent left superior vena cava draining into a dilated coronary sinus. Additional findings included mild tricuspid insufficiency and pulmonary artery systolic pressure of 33mmHg. Biventricular function was adequate, with preserved global and segmental contractility.



Currently, the patient is under surveillance with pediatric cardiology, with regular follow-up visits scheduled at least three times per year. The patient's hemodynamics remain stable, and he is awaiting further evaluation with angiotomography to complement the diagnostic assessment.

This case highlights the importance of comprehensive evaluation and follow-up in patients with congenital venous anomalies such as RSVC agenesis and PLSVC. Multidisciplinary collaboration and ongoing monitoring are essential to ensure optimal management and long-term outcomes for affected individuals.

DISCUSSION

The case presented underscores the clinical significance of right superior vena cava (RSVC) agenesis and persistent left superior vena cava (PLSVC) as rare congenital anomalies with variable clinical presentations. While many patients with these venous anomalies remain asymptomatic, some may present with symptoms related to associated cardiovascular abnormalities or complications.

The presence of RSVC agenesis and PLSVC poses diagnostic challenges, often requiring a multimodal imaging approach for accurate characterization of the venous anatomy. Echocardiography serves as a valuable initial screening tool, with additional imaging modalities such as computed tomography (CT) angiography or magnetic resonance imaging (MRI) providing detailed anatomical information (4, 5).

The presence of symptoms and associated complications primarily guides the management of RSVC agenesis and PLSVC. Conservative measures may be sufficient for asymptomatic patients, while pharmacological interventions or surgical procedures may be necessary for those with symptomatic complications such as thrombosis or arrhythmias (3, 6).

Minimally invasive treatment modalities, such as transvascular closure of persistent left superior vena cava fenestrations, offer promising alternatives to traditional surgical approaches, providing effective closure of anomalous venous pathways with reduced procedural risks (8).

Genetic studies have revealed insights into the underlying pathogenesis of RSVC agenesis and PLSVC, although further research is needed to elucidate the genetic basis and identify potential risk factors for these venous anomalies (13).

Multidisciplinary collaboration among cardiologists, radiologists, and other healthcare providers is essential for comprehensive evaluation and management of patients with RSVC agenesis and PLSVC. Long-term follow-up is crucial to monitor for potential complications and ensure optimal outcomes for affected individuals.

CONCLUSION

In conclusion, RSVC agenesis and PLSVC are rare congenital anomalies of the venous system that present unique challenges in diagnosis and management. While many patients remain asymptomatic, some may develop complications such as thrombosis or arrhythmias, requiring timely intervention.

Advancements in imaging techniques, genetic studies, and minimally invasive treatment modalities have contributed to new perspectives in the understanding and management of these venous anomalies. However, further research is needed to elucidate the genetic basis, pathophysiology, and natural history of RSVC agenesis and PLSVC.

Multidisciplinary collaboration and ongoing monitoring are essential to ensure optimal outcomes for patients with RSVC agenesis and PLSVC. Comprehensive evaluation and follow-up care are necessary to detect and address potential complications and to provide personalized treatment approaches based on individual patient characteristics and clinical presentation.

REFERENCES

Inferior Vena Cava Agenesis: An Underrated Cause of Deep Venous Thrombosis: Kannappan Muthumeena; Sakthi Velavan Sruthilatha. Cureus; Palo Alto Vol. 15, Iss. 1, (2023). https://www.proquest.com/openview/73e7e9ff040651736c91dc54d7f8f3e0/1?pq-origsite=gscholar&cbl=2045583

Prenatal Detection and Postnatal Outcome of Persistent Left Superior Vena Cava and Agenesis of Ductus Venosus Associated with Postnatal Bovine Aortic Arch:

Ceylin Elbistanli , Yılmaz Yozgat , Mehmet Said Dogan , Can Yilmaz Yozgat , Mehmet Serdar Kütük (2023) https://www.thieme-connect.com/products/ejournals/abstract/10.1055/a-2219-9889

Unique Medley of Cardinal Veins: Duplicated Superior and Inferior Venae Cavae With Left Renal Agenesis and Hemiazygos Continuation of Left Inferior Vena Cava With Drainage Into Left Atrium: Selvamurugan Vignesh, and Tameem Ahmad Bhat (2022) https://journals.sagepub.com/doi/abs/10.1177/15385744211051493

Gradual diagnosis and clinical importance of prenatally detected persistent left superior vena cava with absent right superior vena cava – a case report and literature review: Oskar Sylwestrzak and Maria Respondek-Liberska, (2020) https://sciendo.com/article/10.15557/jou.2020.0023

Implantation of a leadless pacemaker in a patient with persistent left superior vena cava and right superior vena cava atresia : Eloisa Mariscal-López, Marta Pombo Jiménez, Almudena Valle Alberca, Francisco Ruiz Mateas, (2022) https://academic.oup.com/europace/article/24/12/1972/6701891

Agenesis of the venous duct with abnormal return of the umbilical vein into the superior vena cava: A case report: Hidehiro Mori MD, Yuki Kawasaki MD, Yosuke Murakami MD, Mitsuhiro Fujino MD, Takeshi Sasaki MD, Kae Nakamura MD, Yoko Yoshida MD, Tsugutoshi Suzuki MD, PhD, Eiji Ehara MD (2022) https://onlinelibrary.wiley.com/doi/abs/10.1111/echo.15423

Double superior vena cava and left brachiocephalic vein agenesis: a rare systemic vein anomaly and potential source of cardiac implantable electronic device and central venous catheter placement complications: R. Steckiewicz, P. Stolarz, M. Marchel, M. Michalak, D. Konecki, E. Szczerba, M. Kowara, L. Grabowska-Derlatka, M. Grabowski https://journals.viamedica.pl/folia_morphologica/article/view/84478

Persistant Left Superior Vena Cava with and Without Right Superior Vena Cava: Significance of Prenatal Diagnosis: Ozge Kahramanoglu, Oya Demirci, Lutfiye Uygur, Nurdan Erol, Antonio Schiattarella & Agnese Maria Chiara Rapisarda (2024) https://link.springer.com/article/10.1007/s00246-023-03353-0

Inferior Vena Cava Agenesis, Bilateral Double Renal Veins, and Left Circumaortic Renal Vein: A Rare Case and Brief Review of the Literature: Karavas, Erdal; Aydin, Sonay; Unver, Edhem; Kadirhan, Ozlem; Kantarci, Mecit (2023) https://journals.lww.com/joai/fulltext/2023/72030/inferior_vena_cava_agenesis,_bilateral_double.15.aspx

Permanent pacemaker implantation in a patient with persistent left superior vena cava with an absent right superior vena cava: A case report: Takeshi Mikami MD (2021) https://www.sciencedirect.com/science/article/pii/S1878540920301729

Persistent left superior vena cava in a 29-year-old lady with Ebstein's anomaly and complete heart block. A case report and literature review:.B.W. Russell, T.R. Koroma, V. Conte, J. Coker, S. Conteh, Kumar Bharat, O.Z. Mahdi (2022) https://www.sciencedirect.com/science/article/pii/S2049080122006446

Absent Right Superior Vena Cava and Persistent Left Superior Vena Cava: An Incidental Finding: Joshi, Swati; Singh, Ajmer: (2023) https://journals.lww.com/aoca/fulltext/2023/26040/absent_right_superior_vena_cava_and_persistent.12.aspx

Persistent left superior vena cava without right superior vena cava during fetal life: Ufuk Ilker Sayici, Mehmet Emre Ari: (2023) https://www.cambridge.org/core/journals/cardiology-in-the-young/article/abs/persistent-left-superior-vena-cava-without-right-superior-vena-cava-during-fetal-life/C0337B56D81C678BF5E0737B59C5C8B3

Isolated left side superior vena cava in a dialysis patient: Right direction, wrong turn! : Sahil Bagai, Dinesh Khullar, Kunal Gandhi, Deepak Panigrahi, Shahnawaz Bashir, Vivek Saxena: (2021) https://onlinelibrary.wiley.com/doi/abs/10.1111/hdi.12990

A case report of prenatal diagnostics and surgical correction of pulmonary agenesis associated with total anomalous pulmonary venous connection and vascular ring: Maksym Delikatnyi, Raad Tammo, Hanna Morkovkina, Oleksandr Teterin, Tetyana Yalynska, Roman Sekelyk: (2023) https://sciendo.com/article/10.2478/rjc-2023-0016