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Case Report

Association of Terminal 6q Deletion Syndrome with Persistent Left Superior Vena Cava Drainage into the Left Atrium: Case Report

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Abstract

Terminal 6q deletion syndrome is an infrequent disorder with dearth of data in the literature. Some reports of chromosome 6q deletion showed an association with congenital heart diseases such as atrial septal defect (ASD), ventricular septal defect (VSD), patent ductus arteriosus (PDA), Tetralogy of Fallot (TOF) and double outlet right ventricle (DORV). Persistent left superior vena cava (PLSVC) drainage into the left atrium (LA) is a rare congenital thoracic venous anomaly. In this paper. We demonstrate the first reported association of 6q deletion syndrome with PLSVC drainage into the LA.

Keywords: Atrial septal defect (ASD), Patent ductus arteriosus (PDA), Tetralogy of Fallot (TOF), Ventricular septal defect (VSD)

INTRODUCTION

Two types of persistent left superior vena cava (PLSVC) are usually detected with cardiac catheterisation diagnostic procedures (Davis et al., 1959). Nearly 50% of patients with PLSVC have other cardiac anomalies, such as Tetralogy of Fallot (TOF), atrial septal defect (ASD), or endocardial cushion defect (Genovese et al., 2021). Furthermore, the presence of PLSVC commonly complicates access to a catheter within the right side of the heart (Goyal et al., 2008).

The first type of PLSVC occurs with an incidence of 0.3% to 2% in the general population and in 4.4% among patients with congenital abnormalities (Genovese et al., 2021; Yousaf and Malak, 2008). The drainage of type 1 PLSVC to the coronary sinus does not cause the mixing of blood, which is considered a non-harmful pathology (Genovese et al., 2021). This is generally asymptomatic when it is not associated with other congenital heart diseases. This is usually detected via cardiovascular imaging as it is performed to investigate other cardiac abnormalities (Goyal et al., 2008; Totorean et al., 2022). In contrast with type 2 PLSVC draining into the left atrium (LA), which has serious consequences and necessitates intervention (Genovese et al., 2021). PLSVC joins the LA via an unroofed coronary sinus, which is the absence of the partition between the coronary sinus and the LA, a direct connection to the LA, or a connection to the left superior pulmonary vein (Genovese et al., 2021). The draining of the PLSVC into the LA anomaly is associated with an increased risk of cyanosis, embolic cerebrovascular stroke, intracerebral abscess, and heart failure (Yousaf and Malak, 2008). The unroofed coronary sinus is one of the rarest ASDs (Davis et al., 1959; Bonardi et al., 2012). It is often affiliated with complex congenital heart disease, primarily heterotaxia syndrome (Bonardi et al., 2012). Prior case reports have shown that 82.2% of PLSVC cases involve right superior vena cava (RSVC) (Çetinarslan et al., 2021).

Case presentation

We present the case of a late preterm (gestational age of 36 weeks) infant girl with a family history of congenital cardiac anomalies. The infant was on room air maintaining SpO_2 80–90 and was diagnosed with hypoxic–ischemic encephalopathy (HIE) stage II. A genetic study showed

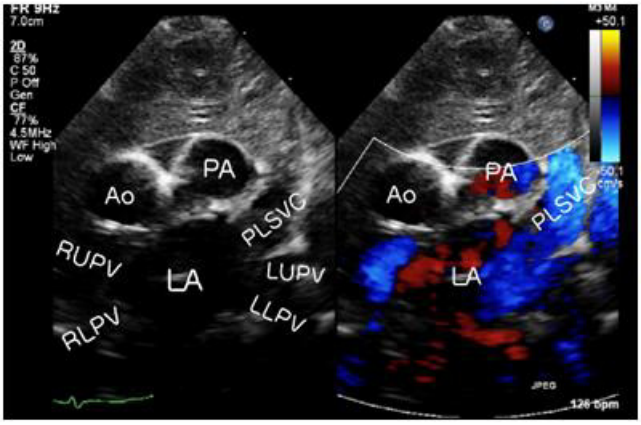
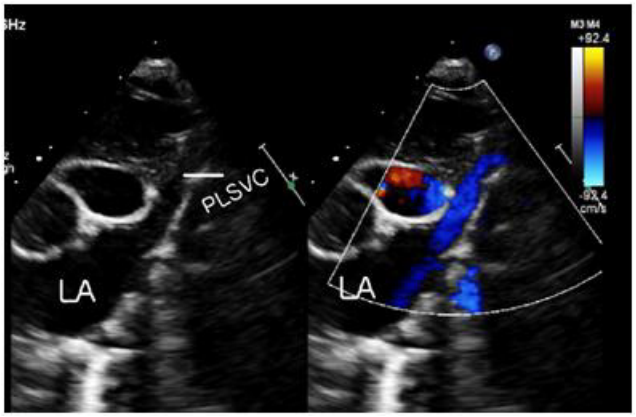
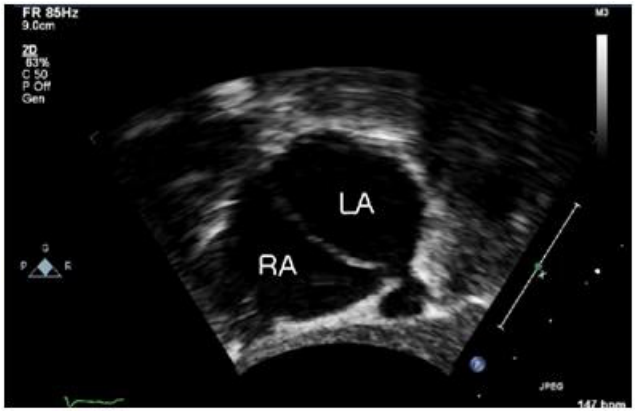


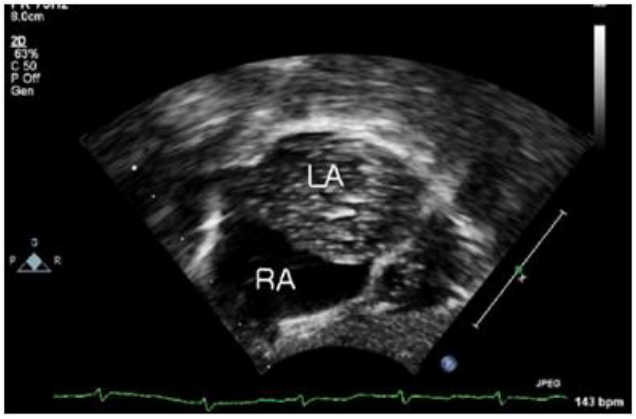
Figure 1. (a) Left side view, Suprasternal notch view of pulmonary veins and left atrium (crab view) with 2D imaging. Right side view, color doppler mapping shows the pulmonary veins and PLSVC draining into the LA. Ao, aorta. PA, pulmonary. RUPV, right upper pulmonary vein. RLPV, right lower pulmonary vein. LUPV, left upper pulmonary vein. LLPV, left lower pulmonary vein. LA, left atrium. PLSVC, persistent left superior vena cava.



(b) Color Doppler flow mapping showing PLSVC draining into the LA.LA, left atrium. PLSVC, persistent left superior vena cava.



(c) Subxiphoid short axis image before bubble study. RA, right atrium. LA, left atrium.



(d). Subxiphiod short axis view after left arm injection with agitated saline (bubble study)showing bubbles entering the LA.LA, left atrium. RA, right atrium.

terminal 6Q deletion. The infant was referred to a pediatric cardiologist to rule out congenital heart disease. Echocardiography was performed revealing PLSVC, patent ductus arteriosus (PDA) and arch flow acceleration. An agitated saline (bubble) study was recommended but not immediately performed. Further echocardiography follow-up was performed with no manifested PDA observed, a patent aortic arch with no coarctation, and PLSVC. A cannula was placed via the subclavian from the left arm. The agitated saline echocardiography (bubble study) was performed and showed PLSVC drainage into the LA (Figure 1).

DISCUSSION

Among congenital anomalies, the manifestation of PLSVC drainage into the coronary sinus is the most common congenital thoracic venous anomaly with a percentage of 0.3–0.5% (3,4,7–9). In 3–10% of patients, the PLSVC drains into the coronary sinus (Al-Muhaya et al., 2020). Rarely, the PLSVC drains into the LA with congenital heart anomalies (Al-Muhaya et al., 2020). In 1800, the first case of PLSVC was reported (Moody et al., 2019).

The venous system begins to form within the fourth week of embryonic life (Bisoyi et al., 2017). The thoracic embryonic venous system includes a pair of superior cardinal veins that return blood from the cranial aspect and inferior cardinal veins, which are responsible for returning the blood from the caudal part (Goyal et al., 2008; Yousaf and Malak, 2008). The right superior and inferior cardinal vein join to form the right common cardinal vein, and the left superior and inferior cardinal vein join to form the left common cardinal vein. An anastomosis between the right and left superior cardinal vein forms the innominate vein at 8 weeks of gestation. The bridging innominate vein may or may not persist. The caudal part of the left superior cardinal vein typically regresses to form the ligament of Marshall. The Persistent caudal part of the left superior cardinal vein performs persistent LSVC (Goyal et al., 2008; Yousaf and Malak, 2008). The types of PLSVC reported in the literature review include double superior vena cava (SVC) without any anastomosis, double SVC with anastomosis, and isolated LSVC with an absent RSVC (Totorean et al., 2022). Diagnosis of PLSVC is mostly an incidental finding discovered during routine investigation for other cardiac abnormalities, performing cardiovascular imaging, invasive procedures, such as cardiac surgery, central line insertion, cardio-thoracic surgery, or complications with catheter insertion (Goyal et al., 2008; Yousaf and Malak, 2008; Bisoyi et al., 2017; Al-Muhaya et al., 2020; Duymus et al., 2012). Generally, it is asymptomatic and has no hemodynamic relevance if not associated with other congenital diseases (Bisoyi et al., 2017; Moody et al., 2019; Duymus et al., 2012). Oxygen desaturation could be absent or infrequent among patients with PLSVC drainage into the LA and bridging vein (Eddine and Kobayashi, 2021). Bridging veins provide interatrial communication (Eddine and Kobayashi, 2021). PLSVC has been reported with the atrioventricular septal defect (VSD), TOF, double outlet left ventricle, double outlet right ventricle, hypoplastic left heart, hypoplastic right heart, pulmonary atresia, single atrium, single ventricle, total anomalous pulmonary venous return, ASD, bicuspid aortic valve, coarctation of the aorta, coronary sinus ostia atresia, and cortriatriatum (Goyal et al., 2008; Totorean et al., 2022). This venous anomaly causes no hemodynamic disturbance (Duymus et al., 2012). However, it has clinical complications (Duymus et al., 2012). PLSVC drainage into the LA occurs right to left shunt, which includes cerebrovascular embolism, cyanosis and heart failure (Yousaf and Malak, 2008).

Additionally, when drugs are applied from the left brachiocephalic, they directly enter the systematic circulation

(Duymus et al., 2012). Furthermore, the effects of PLSVC can involve complications with a permanent pacemaker, implantable cardioverter defibrillator placement and central venous access, cardiopulmonary bypass or catheterisation of the coronary sinus through the PLSVC (3–5,12). Pacemaker implantation is technically challenging and requires skills with cardiac venous drainage anomalies because they are usually unintentionally discovered during intervention (Totorean et al., 2022). The complications can range from simple to more complex (Totorean et al., 2022). It has been reported that the insertion of pacemaker leads or catheters to the PLSVC can result in arrhythmia, cardiogenic shock, cardiac tamponade or coronary sinus thrombosis (Goyal et al., 2008). The last implication is a high incidence of association of PLSVC with congenital anomalies, for example, VSD, ASD, endocardial cushion defect, and TOF (Duymus et al., 2012). The early discovery of this anatomic variant can improve the outcomes for patients (Moody et al., 2019).

Transthoracic echocardiography has a crucial diagnostic role in demonstrating PLSVC (Totorean et al., 2022). However, some deep cardiac structures not clearly delineated using transthoracic echocardiography (Bonardi et al., 2012).

PLSVC without a dilated coronary sinus can increase the suspicion of LA drainage (Aguilar et al., 2019). Other modalities must be performed to resolve any persistent diagnostic doubts (Aguilar et al., 2019), for example, contrast echocardiography, echocardiography with air bubble study, computerized tomography (CT) or magnetic resonance (Aguilar et al., 2019).

Our study used the agitated saline technique, which was injected into the left arm. Echocardiography views revealed bubbles in the LA before the right atrium (RA), which confirmed the connection between the PLSVC and LA.

In 2008, a 72-year-old man with acute renal failure was presented for repair. A left internal jugular haemodialysis catheter was placed. A chest radiograph revealed an unusual course of the catheter. Cardiac CT was performed, demonstrating PLSVC drainage into the LA (Yousaf and Malak, 2008).

In 2012, a 56-year-old patient with rheumatoid arthritis and pulmonary hypertension was presented. Cardiac computer tomography (CT demonstrated bilateral SVC, right SVC with drainage into the RA, and LSVC drainage into the LA. Via an unroofed coronary sinus without communication between the veins (Bonardi et al., 2012).

Furthermore, in 2012, a 69-year-old woman who complained of a cough was admitted to the hospital. Chest CT revealed isolated PLSVC drainage to the LA with RSVC absence and no additional cardiac anomalies (Duymus et al., 2012).

In 2022, a 65-year-old patient with no previous medical history was admitted to the hospital due to syncope episodes. The echocardiography study demonstrated a normal LV systolic function of 55%, mild valvular abnormalities, an enlarged LA and no additional findings. According to electrocardiogram (ECG) Holter monitoring, there was an intermittent third-degree atrioventricular block. Permanent pacemaker implantation was considered for the patient. The failure to access the guidance wire inthe brachiocephalic vein and SVC had caused the suspecting of complex thoracic venous anomaly. The CT showed a hypoplastic SVC, right-sided collaterals with significant tortuosity, and abnormal drainage of dilated tortuous persistent SVC into the inferior pulmonary vein and subsequently into the LA (Totorean et al., 2022).

In our case, the patient was diagnosed with terminal6q deletion syndrome. Terminal 6q deletion syndrome is a sporadic syndrome with limited documented data (Engwerda et al., 2018). Deletion in the 6q chromosome leads to congenital malformation, development delay, or mental retardation (Nair et al., 2012). Some previous patients with chromosome 6q deletion had been associated with congenital heart diseases. Heart anomalies that have been reported include ASD, VSD, PDA, TOF and double outlet right ventricle (DORV) (Boy et al., 1998). In our case, an echocardiography study revealed a PLSVC and normal size of the coronary sinus with no other cardiac congenital anomalies. The agitated saline study confirmed the PLSVC drainage into the LA.

The case presented here is the first report of PLSVC into the LA with no other cardiac anomalies with terminal q6 syndrome.

CONCLUSION

Our case study reveals two important points. First, this report documents the first association in the literature between the rare terminal 6q deletion syndrome and the rare anomaly of PLSVC into the LA. Second, it highlights the importance of agitated saline contrast echocardiography as a mandatory tool to confirm the presence of PLSVC with drainage into the LA. Upon echocardiography, PLSVC with normal coronary sinus should alert the possibility of PLSVC drainage into the LA. PLSVC drainage into the LA causes a right to left shunt, which can result in cerebral vascular embolism, cyanosis and heart failure. Physicians and healthcare providers should be aware of this anomaly, the complications different clinical entities, and variations to improve the outcomes and avoid any possible complications.

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