Diagnosis of absent right superior vena cava with intraoperative transesophageal echocardiography in a child with Holt-Oram syndrome: Anesthetic and perfusion implications

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ABSTRACT
Holt-Oram syndrome is a rare autosomal disorder with cardiac, vascular, and upper limb anomalies. Previous reports have described anesthetic and perioperative challenges including difficulty in arterial and venous cannulations, airway management and rhythm, and temperature abnormalities. There are no previous reports of absent right superior vena cava (SVC) in children with Holt-Oram syndrome. We present images of a case where the diagnosis of absent right SVC with persistent left SVC was made with intraoperative transesophageal echocardiography and discuss the anesthetic and perfusion implications of such findings.

Keywords: Holt-Oram syndrome, transesophageal echocardiography, venous anomalies

INTRODUCTION
Holt-Oram syndrome also called the “heart-hand” syndrome, is a rare autosomal dominant disorder with a prevalence of 1 in 100,000 children.1,2 Upper limb abnormalities in this disorder range from absent thumb to phocomelia. Common cardiac abnormalities include atrial septal defect (ASD), ventricular septal defect (VSD), and conduction abnormalities.3 Vascular anomalies have also been described and include absent or hypoplastic peripheral arteries and veins which has precluded cannulation.1

Intraoperative transesophageal echocardiography (TEE) may be an invaluable modality in such patients presenting for cardiac surgery with the potential to detect additional cardiac and vascular anomalies that may be useful in guiding the surgeons.

CASE REPORT
A 4-year-old male child was referred for preanesthetic evaluation for surgical closure of ostium secundum ASD. The child was small for his age and weighed 10 kg. He had a tall bossing forehead, depressed temples, orbital...
hypertelorism, flat nasal bridge, low set ears, and missing thumbs on both hands [Figure 1a-c]. Mouth opening was restricted to 1.5 fingers. Electrocardiography (ECG) showed right bundle branch block with right axis deviation. Other biochemical parameters were within normal limits. He was referred for a genetic study and returned with a diagnosis of Holt-Oram syndrome.

Anesthesia induction was uneventful including endotracheal intubation on the day of the open-heart surgery. Cannulations included peripheral i.v cannula in the left-hand vein, arterial cannula in the right radial artery and central venous catheter (CVC) in the left internal jugular vein. Prebypass TEE examination was carried out with a pediatric 9T probe (Vivid E95, GE, Vingmed Ultrasound, Horten, Norway) to confirm the ostium secundum ASD and look for any associated anomalies. Ostium secundum ASD measuring 16 mm with a left to right shunt, moderate tricuspid regurgitation and dilated right atrium (RA), and right ventricle were confirmed in standard mid-esophageal (ME) views. However, 2-dimensional (2D) interrogation of the bi-caval view showed a band of fibrous tissue in place of the superior vena cava (SVC) without any evidence of flow [Figure 2a]. Closer inspection of the RA revealed a dilated coronary sinus (CS). An agitated 20 mL saline solution was pushed rapidly through the CVC and contrast enhancement was first seen in the CS instead of RA [Figure 2b and Video loop 1]. ME 4-chamber view with probe turned to left revealed a left SVC which showed similar contrast enhancement to a repeat bolus of agitated saline [Video loop 1]. The findings of absent right SVC were confirmed from the modified deep transgastric bi-caval view when the inferior vena cava (IVC) could be seen with absent right SVC [Figure 2c,d and Video loop 2]. No other cardiac anomalies were detected. The findings were communicated to the surgeons, which were confirmed following median sternotomy. Accordingly, the plan of bi-caval cannulation for establishing cardiopulmonary bypass (CPB) was changed to IVC and left SVC cannulation. Further course was uneventful. Postoperative chest X-ray revealed the CVC along the left heart border [Figure 3].

DISCUSSION

Holt-Oram syndrome is associated with several cardiac lesions including ostium primum and secundum ASDs, VSDs, tetralogy of Fallot, complete atrioventricular canal defects, coarctation of the aorta, endocardial cushion defects, isolated left SVC, etc.[4] Isolated left SVC is when there is agenesis or absence of right SVC and persistence of left SVC. The incidence of left SVC is 0.3–0.5% of the general population and results due to the failure of closure of the left anterior cardinal vein.[5] However, the occurrence of isolated left SVC is extremely rare with an incidence of 0.09–0.13% of patients with congenital heart disease.[6] More than 50% of cases of isolated left SVC are associated with congenital cardiac anomalies like ASDs, endocardial cushion defects, and tetralogy of Fallot.[6] Other situations where the right SVC is absent in its usual location includes children who have previously undergone the Warden’s procedure where the right SVC is anastomosed to the RA appendage or the Glen procedure where the right SVC is anastomosed to the right pulmonary artery.

Various implications of isolated left SVC have been described previously and include difficulty in the insertion of CVC and pulmonary artery catheters, implantable pacemaker leads, arrhythmias, and conduction blockade secondary to dilated CS and right to left shunt in case of unroofed CS or direct drainage of the left SVC into the left atrium.[6] Left ventricular inflow obstruction has also been reported due to left SVC with dilated CS and may even mimic features of cor triatriatum sinister.[7]

In this index case, the diagnosis of absent right SVC was confirmed using the modified deep transgastric bi-caval view. This view has been described previously and is a

Figure 1: (a) Syndromic facies with the tall bossing forehead, orbital hypertelorism, flat nasal bridge, depressed temples, and low set ears. (b and c) Absent thumb
useful view to measure gradients across the SVC since the flows are parallel to the ultrasound beam.[8] Besides, the imaging window for the SVC and IVC is through the liver and so, 2D views can delineate a greater length of both the caval vessels in contrast to the ME bi-caval view where the air-filled right bronchus may obscure the views of the SVC. In reaching a diagnosis of absent caval vessels, both spectral and color flow Doppler (CFD) should be used since there may be artifacts with CFD, especially with low aliasing velocities. Agitated saline bolus to left-hand vein confirms the diagnosis of left SVC where opacification occurs first in the left SVC and CS instead of RA. In the case of absent right SVC, the opacification of the CS before RA will occur even with a right-hand vein injection.

The prebypass TEE diagnosis of persistent left SVC and absent right SVC had implications for the conduct of CPB in this case. It saved critical surgical time, which otherwise would have been spent in searching for the vein by the surgeon after median sternotomy. Additional implications of left SVC are that cannulation is technically more challenging and it also, essentially precludes retrograde administration of cardioplegia for myocardial protection. Finally, using the same central vein for both the CVC as well as the CPB venous drainage cannula may lead to knotting, coiling, or kinking of the CVC.[9]

Diverse problems including the absence of radial artery necessitating the use of alternate arterial cannulation for invasive blood pressure monitoring,[9] significant arrhythmias,[10] and even malignant hyperthermia like manifestations,[11] have all been described in patients with Holt-Oram syndrome. Besides, the possibility of intrathoracic systemic venous anomaly must be kept in mind,[12] which may have both anesthetic and surgical implications in the conduct of cardiac surgery.

In summary, multimodal imaging may be needed to manage such complex cases, including the use of ultrasound for peripheral vascular and central venous cannulations and echocardiography for diagnosis of cardiac and vascular abnormalities.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

REFERENCES
1. Basson CT, Cowley GS, Solomon SD, Weissman B, Poznanski AK, Traill TA, et al. The clinical and genetic spectrum of the Holt-Oram syndrome (heart-hand syndrome). N Engl J Med 1994;330:885-91.
2. Huang T. Current advances in Holt-Oram syndrome. Curr Opin Pediatr 2002;14:691-5.
3. Singh A, Pathania VS, Girotra S, Iyer KS. Anesthetic implications in Holt-Oram Syndrome. Ann Card Anaesth 2013;16:157-8.
4. Spiridon MR, Petris AO, Gorduza EV, Petras AS, Popescu R, Caba L. Holt-Oram syndrome with multiple cardiac abnormalities. Cardiol Res 2018;9:324-9.
5. Fisher DR, Zuberbuhler JR. Anomalous systemic venous return. In: Anderson RH, Baker E, McCartney RF, editors. Paediatric Cardiology.
New York: Churchill Livingstone; 2002. p. 851-65.

6. Bisoyi S, Jagannath U, Dash AK, Tripathy S, Mohapatra R, Pattnaik NK, et al. Isolated persistent left superior vena cava: A case report and its clinical implications. Ann Card Anaesth 2017;20:104-7.

7. Vargas FJ, Rozenbaum J, Lopez R, Granja M, De Dios A, Zarlenia B, et al. Surgical approach to left ventricular inflow obstruction due to dilated coronary sinus. Ann Thorac Surg 2006;82:191-6.

8. Aggarwal N, Unnikrishnan KP, Sunel PR, Mathew T. Modified deep transgastric bicaval view for revealing superior vena caval obstruction in a patient undergoing sinus venous atrial septal defect repair: A case report. J Cardiothorac Vasc Anesth 2016;30:729-32.

9. Misra S, Sinha PK. An unusual case of looping of the central venous catheter: Who is the culprit? J Cardiothorac Vasc Anesth 2008;22:336-7.

10. Rana M, Solanki SL, Agarwal V, Divatia JV. Holt-Oram syndrome: Anesthetic challenges and safe outcome. Ann Card Anaesth 2017;20:110-1.

11. Franklin AD, Lorinc AN, Donahue BS. Malignant hyperthermia-like manifestations in a two-month-old child with Holt-Oram syndrome undergoing cardiac surgery. J Cardiothorac Vasc Anesth 2014;28:1326-7.

12. Varma PK, Padmakumar R, Hari Krishnan S, Koshi T, Neelakandan KS. Holt-Oram syndrome with hemiazygous continuation of inferior vena cava. Asian Cardiovasc Thorac Ann 2006;14:161-3.