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Isolated Right Superior Vena Cava Drained to Left Atrium in a Child with Waardenburg Syndrome and Neurofibromatosis Type I

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Abstract

Isolated right superior vena cava (SVC) draining to the left atrium (RSVC-LA) is an extremely rare cyanotic congenital heart disease (CHD). Such lesion is easily missed with improper scanning or inattentive interpretation of echocardiography. This can result in potential systemic severe complications. We report a child with RSVC-LA who has two significant non-cardiac co-morbidities, including Waardenburg syndrome and neurofibromatosis type I (NF1). This patient was referred to cardiology assessment due to unexplained low saturation and was diagnosed as RSVC-LA; however, fortunately not yet showing complications of systemic thromboembolic phenomenon (STEP).

Keywords: Right superior vena cava, Left atrium, Cyanosis, Thromboembolism, Echocardiographic bi-caval view, Waardenburg, Neurofibromatosis

1. Background

Right superior vena cava to left atrium is a rare cyanotic congenital heart disease (CHD) and almost with subtle clinical findings.

Clinical diagnosis is challenging, however the prenatal diagnosis is possible [1]. Post-Nataly, the abnormal SVC drainage seen in the standard bi-caval view in the echocardiography might be the first key for the diagnosis. The confirmation of this diagnosis can be achieved by additional contrast echocardiography and other imaging modalities as computed tomographic angiography (CTA), magnetic resonance imaging, and radionuclide imaging.

The treatment of RSVC-LA is surgical with multiple strategies including, transecting the right SVC from the LA and anastomosing it to the right atrial appendage, patching the right SVC to the right atrium (RA), and extra-cardiac approach [2]. There should be a balance between the advantages of early repair to avoid potential risks of STEP complications

versus delaying the repair till optimizing patient condition and stabilizing any significant co-morbidities.

2. Case report

A twenty-nine-month-old boy, delivered at 34 weeks gestation, with a 1.5 kg birth weight. The patient persistently showed low oxygen saturation (85–90%) and recurrent admission for sepsis. Therefore he was referred to our center for further evaluation and management. During admission, we noticed the presence of a few localized hyperpigmentation and some subtle dysmorphic features. The patient had a significant family history of autosomal dominant diseases (mother and older brother diagnosed as neurofibromatosis type 1 and father has Waardenburg syndrome). The patient was on oxygen since birth (2 L/Minute by nasal cannula) and tube feeder due to swallowing dysfunction.

Multidisciplinary evaluations started to evaluate the multiple comorbidities of the patient, including

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the primary team general pediatric for overall care and severe failure to thrive (FTT), genetic team for syndromic diagnosis, neurology team (for neurologic manifestations of neurofibromatosis and Waardenburg syndrome), gastroenterology team (for swallowing dysfunction), pediatric surgery team (for gastrostomy tube insertion), dietician (for nutritional assessment and rehabilitation), and cardiology team (to evaluate for a possibility of cyanotic CHD).

The laboratory findings were within normal range. ECG was unremarkable without signs of ischemia which could be a complication of possible STEP.

Standard transthoracic echocardiography (Fig. 1) revealed RSVC-LA and an atrial septal defect-secundum type (ASD-II) with left to right shunt. We confirmed the diagnosis of RSVC-LA and exclude the possibility of anomalous pulmonary venous drainage by CTA (Fig. 2a,b).

The extensive evaluation for co-morbidities is ongoing with out-patient follow-up. This patient below the third percentile in growth chart, last clinic visit at age of thirty two months where his body weight merely 3.7 kg.

3. Discussion

Right SVC to left atrium is considered an occurrence of abnormal systemic venous connection. It is a rare cyanotic CHD and extremely rare when presented as isolated CHD. Worldwide incidence is less than 0.5% of all CHD [3]. Similar cases were reported in KSA [4,5]; however, the actual national incidence is not known.

Manifestations vary from asymptomatic patients to unrecognized sub-normal saturation to mild

cyanosis and mild clubbing of fingers and toes reaching to serious manifestations of STEP. This could include seizure disorder [6] life-threatening brain abscess, which are not easily attributed to such extremely rare CHD [7]. The variable manifestations in our patient reflect the multiple co-morbidities. Among these findings, the relevant clue was the persistent low saturation that was not normalized by offering the patient oxygen supply.

Laboratory workup showed the effects of infection recovery like high WBC or nutritional deficiencies like low vitamin D level. The screening for other significant abnormalities like endocrine deficiencies, or general indicators of ongoing systemic inflammatory disorder was unremarkable. Genetic testing revealed a positive gene for Waardenburg disease. NF1 was diagnosed clinically by typical clinical criteria and positive family history.

The diagnosis of RSVC-LA usually depends on the high suspicion of the physician during evaluation for the rare cause of cyanotic CHD and a well-standardized approach during transthoracic echocardiographic evaluation, especially in the bi-caval view. For more confirmation, contrast echocardiography [8,9] and or CT angiography [10] can play an important role. The brain MRI of our patient was reported as normal.

Treatment of RSVC-LA is a surgical repair that must be performed as early as possible to prevent paradoxical embolization and the risk of brain abscesses [3]. The significant co-morbidities in our patient like severe and poorly respond FTT, recurrent pulmonary infection, and presence of two

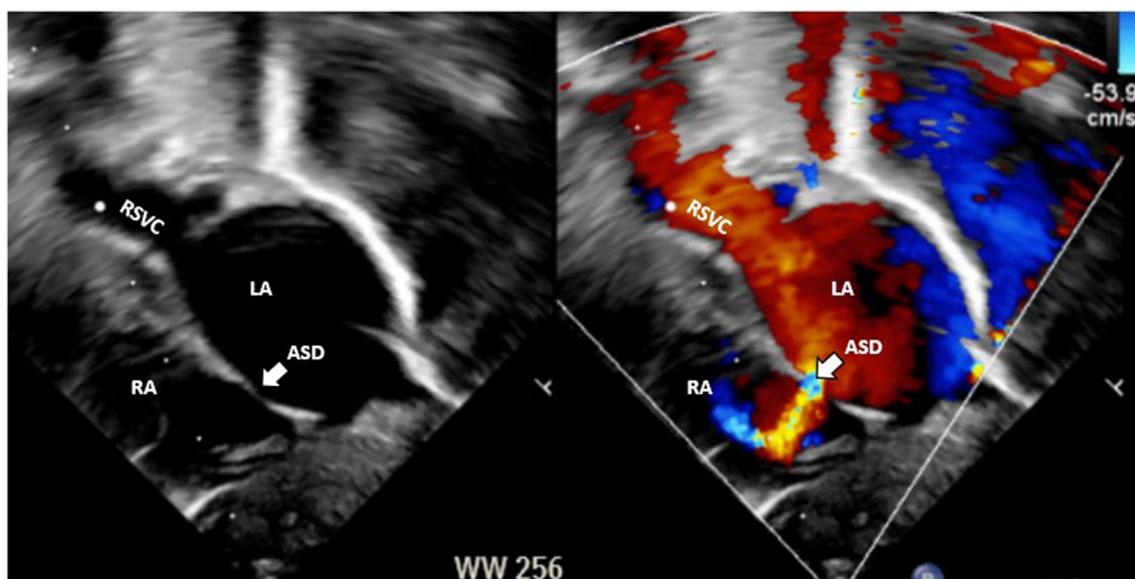


Fig. 1. Subcostal echocardiographic view with color comparison showed RSVC-LA. RA = right atrium, ASD = atrial septal defect, arrow.

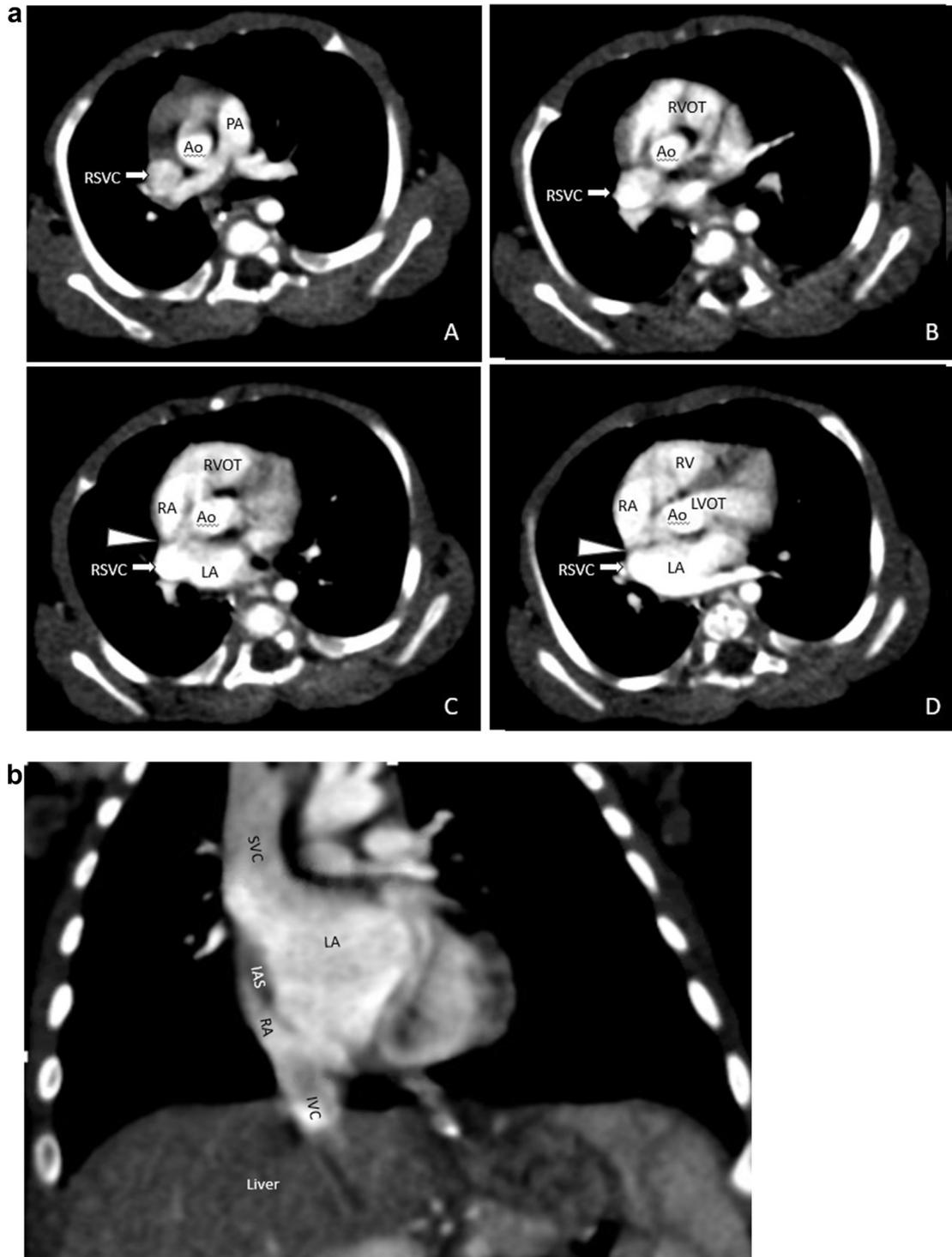


Fig. 2. a: Computed tomographic angiographic scanning. Axial views arranged cephalo-caudal A to D showed Right Superior Vena Cava (RSVC) drained to dilated left atrium (LA). Ao = Aorta, LVOT = left ventricle outflow tract, PA = pulmonary artery, RV = right ventricle, RVOT = right ventricle outflow tract, Arrow head = interatrial septum. b: Computed tomographic angiographic reformate scanning. Coronal view RSVC-LA. IAS = interatrial septum, IVC = inferior vena cava, RA = right atrium.

autosomal dominant diseases, resulted in postponing the surgical correction. The surgeon preferred to optimize the patient's condition before

offering him surgical repair, therefore the patient was not yet operated till time of the submission of this case-report.

4. Conclusion

The current case presented a rare combination of RSVC-LA with two rare dominant syndromes, a genetically proven Waardenburg syndrome and clinically diagnosed Neurofibromatosis type I. As far as we know, such combination is the first reported case in the literature. Although early surgical repair is the standard management to avoid the potential risk of systemic thromboembolism, it deferred in our case due to un-resolved significant co-morbidities.

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Author contribution

Conception and design of Study: OAA, YHA. Literature review: OAA. Acquisition of data; OAA. Analysis and interpretation of data: OAA. Data collection; Drafting of manuscript: OAA. Revising and editing the manuscript critically for important intellectual contents: YHA, ASA. Data preparation and presentation: OAA. Supervision of the research: YHA, ASA. Research coordination and management: OAA, YHA, ASA.

Conflict of interest

The authors declared no potential conflicts of interest concerning the research, authorship, and/or publication of this article.

Ethical issue

The Ethics Committee of KFMC approved our case report.

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