Case Report

Situs ambiguous with three chambered heart (single atrium), evaluation by four-dimensional X-strain color echocardiography – A case report

Akhil Mehrotra[1], Ajay Sharma[2], Swati Singh[2], Shubham Kacker[3]

From [1]Chief, Pediatric and Adult Cardiology, [2]Senior Echocardiography Technician, Prakash Heart Station, Nirala Nagar, Lucknow, Uttar Pradesh, [3]Senior Coordinator, Project Management Office, Tech Mahindra, New Delhi, India

ABSTRACT

Situs ambiguous accounts for 4% of all congenital heart disease (CHD) and has an incidence of 1:10,000 new born births. To standardize the nomenclature for CHD, the international nomenclature society published a globally accepted nomenclature tree for CHD with the 11th Iteration of international classification of disease. According to this publication common atrium, single atrium and atrium communis are denoted as synonyms. The incidence of common atrium among atrial septal defect (ASD) patients is 3–4% only and thus denoting it to be a rare entity. We are presenting an exceedingly rare case of a patient of situs ambiguous, left isomerism, and single atrium with a cleft on the anterior mitral leaflet, evaluated comprehensively by four-dimensional X-strain color Doppler echocardiography.

Key words: Heterotaxy, Levocardia with situs ambiguous, Single atrium, Situs ambiguous

Situs ambiguous or heterotaxy syndrome refers to an abnormal positioning of internal organs relative to the normal [1]. Situs Ambiguous has an incidence of 1:10,000 newborn births, but accounts for about 4% of all the congenital heart disease (CHD) [2]. The left isomerism or situs ambiguous with polysplenia has an incidence of between 1:10,000 and 1:20,000 newborn births with a female preponderance [3]. Although the term polysplenia is used, the number of spleens ranges from one to ten or more, and they may be located either bilaterally or, if unilateral, ipsilateral to the stomach (Fig. 1) [3].

The terms common atrium (CA) and single atrium (SA) have been used interchangeably in the literature. CA was first reported by Young and Robinson in 1907 [4]. In 1959, Ellis *et al.* published an article about the diagnosis and surgical treatment of CA (Cor triloculare biventriculare) [5]. The term "CA" is used for the condition, where there is (i) either atrial septum is completely absent or it is represented by the small strand of tissue at the superior atrial wall of the common chamber, (ii) no interventricular communication, and (iii) atrioventricular (AV) cushion defect [6]. A cleft mitral leaflet is almost always present with this condition [7].

Levy *et al.* described SA (also known as cor triloculare biventriculare) as one of the rare congenital anomaly identified by the complete absence of the atrial septum without an endocardial cushion defect. It is characterized by: (i) complete absent atrial septum, (ii) no AV valves malformation, and (iii) no interventricular communication. They suggested that the term CA should be used to denote the condition of complete absence of the atrial septum, accompanied by malformation of the AV valves, with or without interventricular communication [8].

Therefore to standardize the nomenclature for CHD across the globe, the International Nomenclature Society linked its efforts with those of the World Health Organization, to obtain a globally accepted nomenclature tree for CHD with the 11th Iteration of the International classification of diseases. According to this publication [9], common atrium with separate AV junctions has been defined as "a congenital cardiovascular malformation, in which there is complete or near-complete absence of the Interatrial septum" and single atrium and atrium communis are denoted as synonyms.

Atrial septal defect (ASD) occurs in 1 in 1500 live births, accounting for 10–15% of congenital heart defects in children and the incidence of common atrium among ASD patients is 3–4% only [10]. In necropsy studies of the congenital heart, the common atrium represented only 0.3% of the cases, and in approximately 2% of the specimens of ASD, there was a complete absence of the atrial septum (Fig. 2) [6].

Echocardiography is usually the first step in diagnosing single atrium and is extremely helpful and beneficial, in the evaluation of coexistent congenital abnormalities [11].

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Correspondence to: Akhil Mehrotra, Chief, Non-Invasive Cardiologist, Pediatric and Adult Cardiology, Prakash Heart Station, D-16, Nirala Nagar, Lucknow, Uttar Pradesh, India. E-mail: sadhnamehrotra14@gmail.com

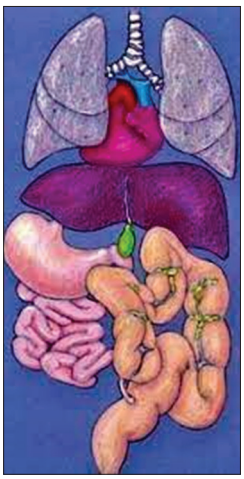

Figure 1: Drawing of situs ambiguous showing bilateral trilobed lung, left-sided liver, and right-sided stomach

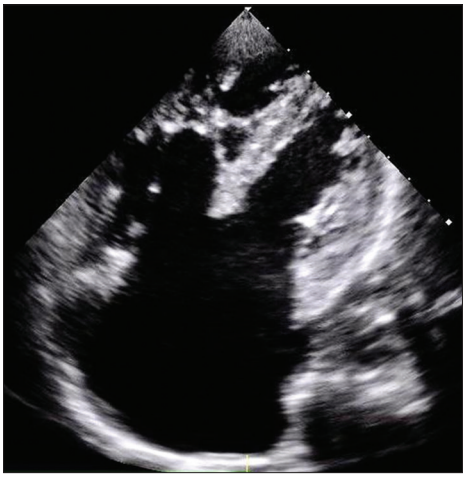

Figure 2: Single atrium in apical 4 ch view (in diastole)

Echocardiography has substantially expanded the horizons, for accurate non-invasive diagnosis of congenital cardiac defects including complex CHDs. Color Doppler echocardiography [12] not only provides additional information, but also improves overall specificity of two-dimensional (2-D) echocardiography. In current clinical practice, the most often used non-invasive imaging method is echocardiography for sequential analysis of complex CHD. Echocardiography is a non-invasive imaging method that can be performed bedside, no radiation exposure occurs, it is an inexpensive method, and it can be repeated at any time [13]. Transesopheged (TEE) and transthoracic echocardiography (TTE) are reliable in diagnosing most CHD in the pediatric and adult populations [14,15]. Additional imaging is required if the goals of imaging are not achieved with echocardiography. Most often non-invasive imaging options include, cardiac magnetic resonance imaging, and cardiac computed tomography (CT) [16].

We are presenting an exceedingly rare case report of situs ambiguous with three chambered heart (single atrium), comprehensively evaluated by four-dimensional X-strain echocardiography.

CASE REPORT

A 5-year-old female child was referred to us from a private pediatric hospital, to rule out the presence of cyanotic CHD. The child was full term delivery by cesarean section, had a normal birth weight, and was delivered at a district hospital, from a primipara woman of 19 year of age. The vaccination of mother and the child was appropriately carried out at the date of presentation to us. There was no history of maternal risk factors of CHD (morbid obesity, diabetes, febrile illness, smoking, alcohol intake, teratogenic drug use, or radiation exposure) on interrogation; her parents gave history of presence of cyanosis since birth, along with recurrent chest infections, failure to thrive and shortness of breath on mild effort. On clinical examination, the child was found to be thin built, irritable, and apprehensive and had frequent bouts of crying while being examined. She had a weight and height of 10 kg and 60 cm respectively, pulse rate was 78/mm, blood pressure was 86/60 mm Hg, and SPO_2 was 75% at room air. The child was deeply cyanosed with bluish coloration of tongue, lips, all the fingers, and toes, together with clubbing. There was a typical pectus carinatum deformity of the chest without any other musculoskeletal anomalies. All the peripheral pulses were normally palpable without any radio femoral delay. Meanwhile, central nervous system, respiratory system, and abdomen were also examined and no abnormality was detected.

On cardiovascular examination, cardiac apex was shifted in the left axillary line, suggestive of cardiomegaly. There was presence of Grade 3/6 pansystolic murmur over left ventricular (LV) apex, radiating to the left sternal edge, and another Grade 3/6 pansystolic murmur was heard at the lower left sternal border, which increased with inspiration, indicative of presence of moderate-to-severe mitral regurgitation (MR), and tricuspid regurgitation (TR), respectively. The second heart sound was closely split and the P2 component was loud. X-ray chest PA view was showing marked cardiomegaly occupying nearly 75% of mediastinum. Moreover, the main and branch pulmonary arteries are strikingly dilated with increased pulmonary blood flow. On more closely scrutinizing the chest X-ray, we found gastric bubble to be on the right side, suggestive of situs inversus/situs ambiguous (Fig. 3).

Hence, a comprehensive abdominal ultrasound was carried out by Chief Radiologist of our institution. The ultrasound revealed number of important salient features: (a) Situs ambiguous (b) liver parenchyma was normal, and occupying the midline and left hypochondrium, (c) stomach is on the right side, (d) there is presence of polysplenia, as evidenced by multiple spleens (3–4), being visualized on the right side along the greater curvature of stomach, (e) both kidneys are normal in position and size, and (f) aorta is on the right side and inferior vena cava (IVC) is on the left (Fig. 4a). On examining the ECG (Fig. 4b), there was normal sinus rhythm, left axis deviation, and Notable Katz-Wachtel phenomenon in the precordial leads, suggestive of biventricular hypertrophy.

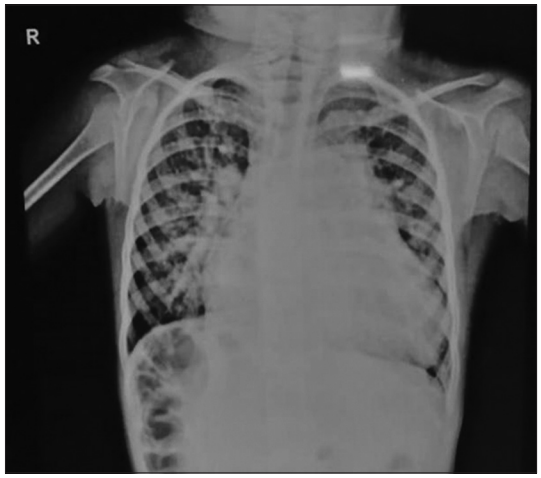

Figure 3: X-ray chest PA – shows massive cardiomegaly with enlarged main and branch pulmonary arteries, suggestive of increase pulmonary blood flow

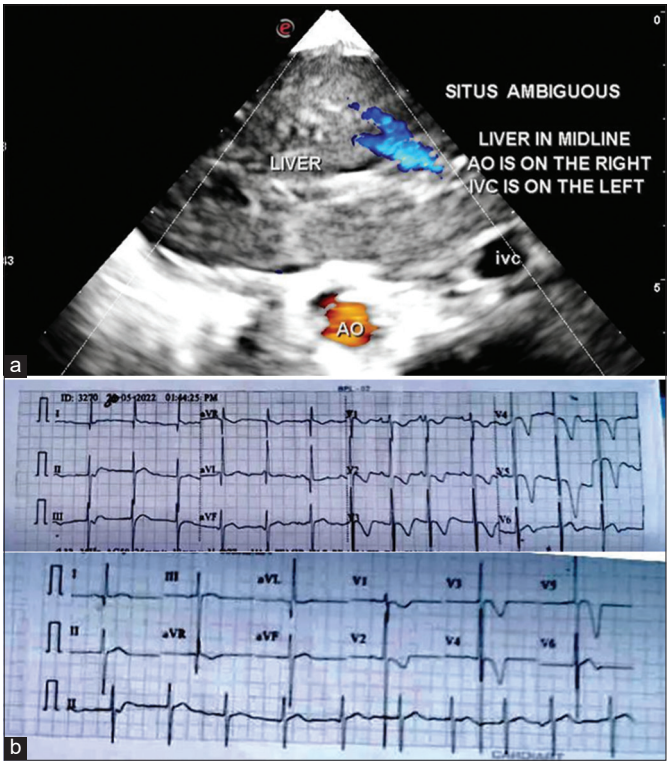

Figure 4: (a) Subcostal view – There is situs ambiguous with liver lying transversely from midline to left hypochondrium, aorta is on the right side and IVC is on the left. (b) ECG suggestive of – left axis deviation with Katz-Wachtel phenomenon characteristic of Biventricular hypertrophy

Comprehensive four-Dimensional X-Strain Echocardiography

The patient underwent four-dimensional X-strain color echocardiography in supine and left lateral decubitus posture. There were levocardia, concordant D-loop, left aortic arch, and normally related great arteries with confluent pulmonary arterial system. There was noticeable presence of a prominent dilated single atrium, with total absence of remnants of atrial septal tissue, both at the roof of single atrium and atrioventricular junction (Fig. 5).

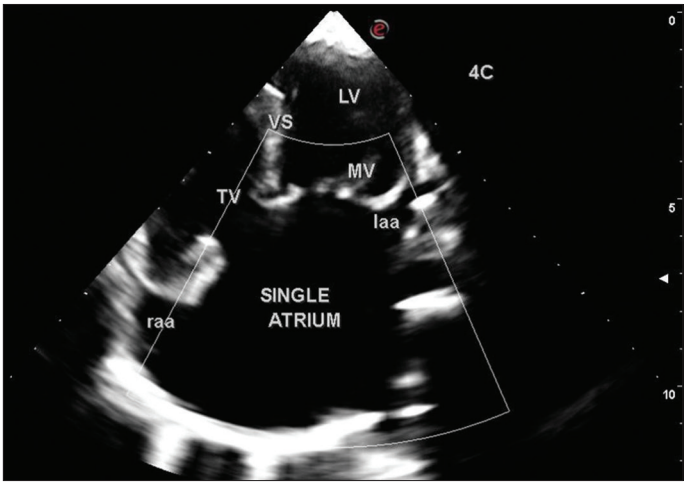

Figure 5: Single atrium with total absence of atrial septal tissue

Superior vena cava (SVC) and IVC were draining into the right lateral side (dextral part) (Fig. 6).

Moreover, three pulmonary veins were opening into the left lateral side (sinistral part) (Fig. 7) of single atrium. The single atrium connects with the left and right ventricle through bileaflet mitral valve (MV) and trileaflet tricuspid valve (TV), respectively (Fig. 8).

Identical left and right atrial appendages were also discerned. Mitral leaflets are large (LAX view) (Fig. 9), with presence of a conspicuous cleft on the anterior mitral leaflet (SAX view) (Fig. 10).

The chordae and papillary muscle of MV and TV were normally attached to the wall of LV and to the ventricular septum and RV apex, respectively.

A classical goose neck deformity was also detected (Fig. 11).

There was presence of severe MR, filling up nearly half of single atrium. Two distinct jets of MR were viewed (Fig. 12).

MR jet 1 was produced due to the cleft in the anterior mitral leaflet while, MR jet 2 was due to incomplete coaptation of mitral valve. Immediately after the emergence of MR jets from mitral valve, they merged with each other to produce a large MR jet image in the cavity of single atrium. Total MR jet area was 16.05 sq. cm. (Fig. 13).

Meanwhile, moderately severe TR was also visualized, with a normal tricuspid valve and a dilated tricuspid valve annulus, with an estimated right ventricular systolic pressure/pulmonary arterial pressure to be 59 mmHg. Moreover, there was moderate biventricular dilatation with normal biventricular systolic function; LV ejection fraction was 67%.

DISCUSSION

Situs ambiguous is an extremely rare entity, occurring roughly 1/40,000 live births, with a male: female ratio of 2:1 [17]. It does not follow a fixed pattern in all the cases [18]. There are two major clinical sub-divisions of situs ambiguous: (a) Left isomerism and (b) right isomerism. Left isomerism consists of situs ambiguous, polysplenia, two bilateral lung lobes, and identical right and left atrium. Right isomerism comprises situs ambiguous, asplenia, and

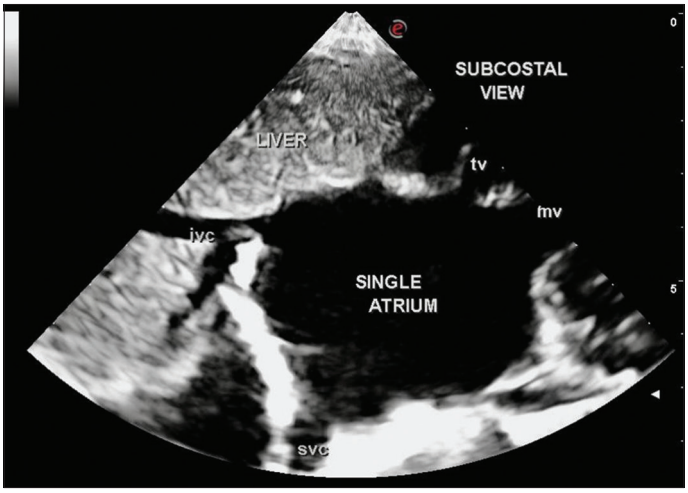

Figure 6: Subcostal view – inferior vena cava and superior vena cava entering the single atrium on the right lateral side (dextral part)

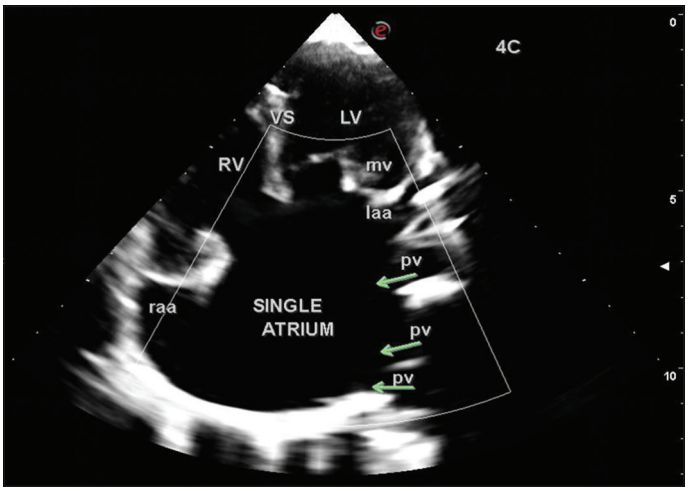

Figure 7: Apical 4 ch view – three pulmonary veins opening into the left lateral side of single atrium (sinistral part). Identical left and right atrial appendages are also visualized

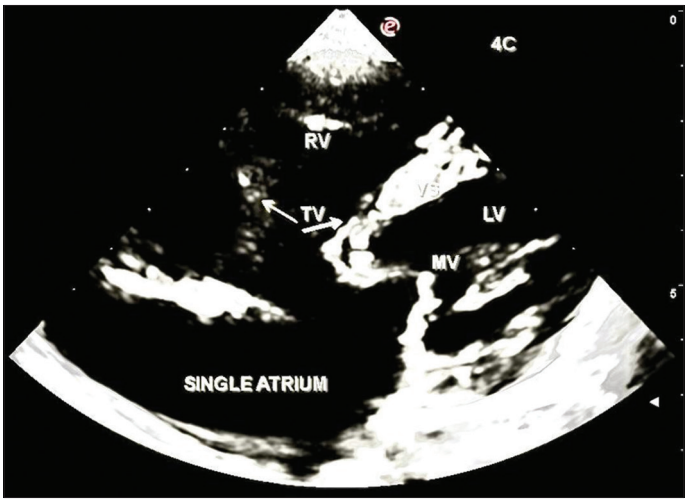

Figure 8: Single atrium apical 4 ch view – single atrium connecting to left and right ventricle through mitral valve and tricuspid valve, respectively

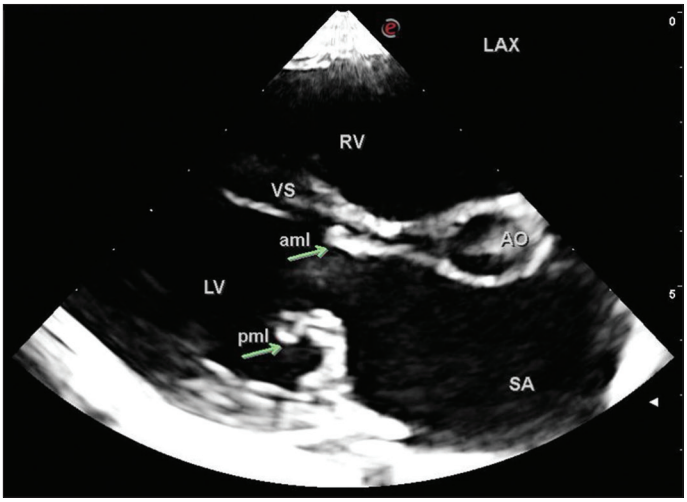

Figure 9: LAX view of single atrium – thickened and large anterior and posterior mitral leaflets

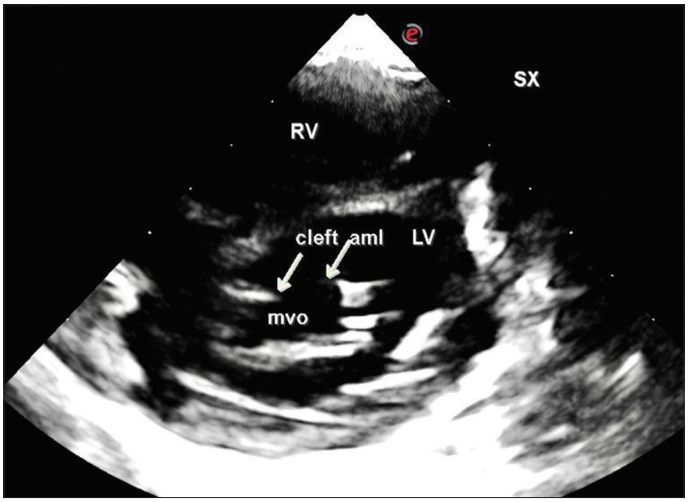

Figure 10: SAX view of mitral valve of single atrium – a conspicuous cleft is seen on the anterior mitral leaflet

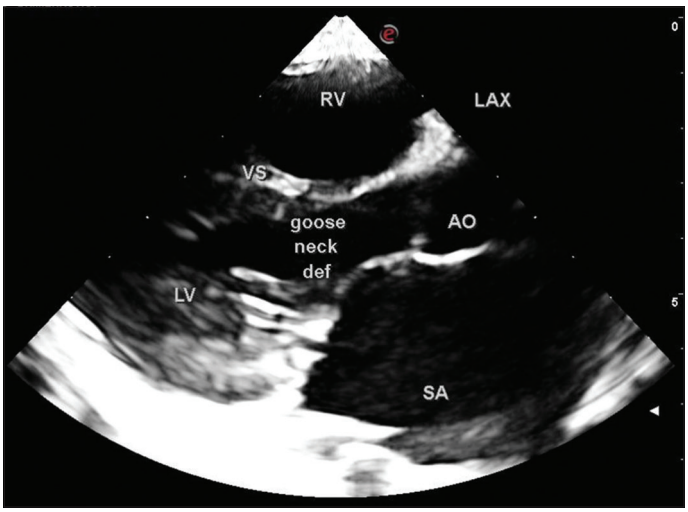

Figure 11: LAX view of Single atrium – classical goose neck deformity present

three bilateral lung lobes [19]. With situs ambiguous, 90–99% of the patients have severe CHD. Our case had situs ambiguous with the left isomerism, in the form of the right-sided stomach, polysplenia, both the atrial appendages being similar in morphology and abdominal aorta was on the right side with IVC on the left.

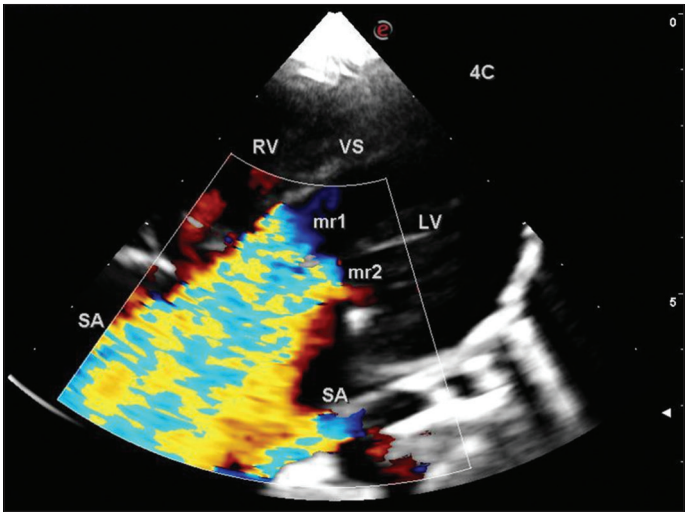

Figure 12: Single atrium – Apical 4 ch view – two distinct jets of mitral regurgitation visualized, which lower down in the left atrium coalesce together

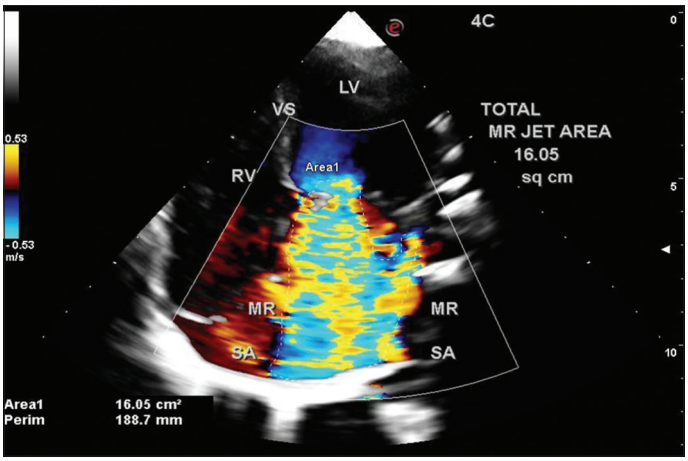

Figure 13: Apical 4 ch view – huge mitral regurgitation jet of size 16.05 sq cm is evident, nearly occupying 50% of single atrial cavity

In single atrium patients, the atrial septum is completely absent. The dextral part of the single atrium has the morphology of a right atrium (crista terminalis, pectinate muscle, and right atrial appendage) and receives the superior and inferior vena cava and coronary sinus. The sinistral portion of the single atrium has the morphology of a left atrium (smooth non-trabeculated walls, a left atrial appendage) and receives the pulmonary veins [7].

Single atrium is usually diagnosed in childhood, though adult presentation of the disease has also been reported. Patients present with decrease in exercise tolerance, shortness of breath, cyanosis, upper respiratory tract infections, fatigue, developmental delay, and heart failure [20]. Single atrium may be associated with different ventricular morphologies: Two ventricles, single right ventricle, single left ventricle, or an undifferentiated single ventricle. Associated malformations include intracardiac anomalies (dextrocardia, mesocardiac, levoverted heart, arial isomerism, complete AV septal defect, and single AV valve) and anomalies of great vessels (anomalous pulmonary venous connection, left-sided SVC, right-sided aortic arch, persistent ductus arteriosus, pulmonary artery anomalies, and aorta pulmonary collaterals) [21]. It is almost always associated with a cleft in the anterior leaflet of the mitral valve [7]. Our patient had single atrium with identical left and right appendages and a cleft on the anterior mitral leaflet.

The hemodynamic features of shunting in single atrium are very similar to those of a large ASD. However, in patients with single atrium, pulmonary and systemic venous blood tend to mix more easily, and systemic arterial oxygen saturation tends to be lower than in those with a large ASD [6]. The natural course of single atrium is poor. Later, identification of single atrium increases the possibility of progression to arrhythmia, pulmonary vascular disease, or cardiac dysfunction [22]. Moreover, surgical correction can considerably slow progression of the disease [23]. Therefore, early diagnosis is important even in well-tolerated cases.

Detailed anatomic evaluation is mainstay for effective patient management. TEE and TTE play a pivotal role in defining the anatomy of the single atrium and ruling out the classical partial AV septal defect; however, it lacks delineation of the extracardiac structures [14,15]. Patients with single atrium without any associated malformations usually have a good prognosis even without surgical management [20]. However, patients with associated malformations usually require surgical intervention [24].

Isolated anterior mitral leaflet clefts are rare, and isolated posterior mitral leaflet clefts are extremely rare, with only a few cases reported in the literature [25]. Recently, a case series reported, two elderly patients with multiple clefts: one has two clefts on the posterior mitral leaflet, and the other one had one each, on the anterior and posterior leaflet [26]. Mitral valve cleft (MVC) is the most common cause of congenital MR [27], and surgical correction might be an effective treatment for patients with MVC, especially for those with moderate-to-severe MR (even if asymptomatic) [24]. In our patients, there was a single cleft on the anterior mitral leaflet, which along with abnormal coaptation of MV, produced a severe MR occupying nearly 50% of single atrium, thus highlighting, that early surgical correction of this complex anomaly should be contemplated at an intensified manner.

CONCLUSION

To the best of our knowledge, this is a first case report of situs ambiguous, let isomerism, single atrium, and cleft on the anterior mitral leaflet, evaluated comprehensively by four-dimensional X-strain color Doppler echocardiography. The presence of severe MR along with intense cyanosis and profound shortness of breath warranted an accelerated action for early surgical correction.

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REFERENCES

1. Applegate V, Goske MJ, Pierce G, Murphy D. Situs revisted: Imaging of the heterotaxy syndrome. Radiographics 1999;19:837-54.
2. Paladini D, Volpe P. Ultrasound of Congenital Fetal Anomalies: Differential Diagnosis and Prognostic Indicators. London, United Kingdom: Informa; 2007.
3. Duran MA, Guerena A. Sequence of right laterality with spleen: Widening the spectrum of heterotaxy. Pediatr Pathol Mol Med 2002;21:461-5.
4. Young AH, Robinson A. Some malformation of the human heart. M Chron 1908;47:96-106.
5. Ellis FH Jr., Kirklin JW, Swan HJ, Dushane JW, Edwards JE. Diagnosis and surgical treatment of common atrium (cor triloculare-biventricular). Surgery 1959;45:160-72.
6. Rastelli GC, Rahimtoola SH, Ongley PA, McGoon DC. Common atrium: Anatomy hemodynamics, and surgery. J Thorac Cardiovasc Surg 1968;55:834-41.
7. Munoz-Armas S, Gorrin JR, Anselmi G, Hernandez PB, Anselmi A. Single atrium. Embryologic, anatomic, electrocardiographic and other diagnostic features. Am J Cardiol 1968;21:639-52.
8. Levy MJ, Salomon J, Vidne BA. Correction of single and common atrium, with reference to simplified terminology. Chest 1974;66:444-6.
9. Franklin RC, Beland MJ, Colan S, Walters HL, Aieollo VD, Anderson RH, *et al*. Nomenclature for congenital and pediatric cardiac disease: The international pediatric and congenital cardiac (IPCCC) and the eleventh iteration of the international classification of disease (ICD-11). Cardiol Young 2017;27:1872-938.
10. Van der Linde D, Helbing AW, Takkenberg JM, Ross-Hesselink WJ, Konings EM, Slager AM, *et al*. Birth prevelance of congenital heart disease worldwide: A systemic review and meta-analysis. J Am Coll Cardiol 2011;58:2241-7.
11. Demirelli S, Firtina S, Ermis E, Inci S. Common Atrium a Rare Congenital Heart Anomaly. Turk Kardiyoloji Dernegi Arsivi-Archives of the Turkish Society of Cardiology; 2015. p. 579.
12. Huhta JC, Hagler DJ, Seward JB, Tajik AJ, Jnlsrud PR, Ritter DG. Two-dimensional echocardiographic assessment of dextrocardia: A segmental approach. AM J Cardiol 1982;50:1351-60.
13. Havasi K, Ambrus N, Kalapos A, Forster T, Nemes A. The role of echocardiography in the management of adult patients with congenital heart disease following operative treatment. Cardiovasc Diagn Ther 2018;8:771-9.
14. Dorfman AL, Levine JC, Colan SD, Geva T. Accuracy of echocardiography in low birth weight infants with congenital heart disease. Pediatrics 2005;115:102-7.
15. Tworetzky W, McElhinney DB, Brook MM, Reddy VM, Hanley FL, Silverman NH. Echocardiographic diagnosis alone for the complete repair of major congenital heart defects. J Am Coll Cardiol 1999;33:228-33.
16. Hiratzka LF, Bakris GL, Beckman JA, Bersin MR, Carr FV, Casey ED Jr., *et al*. 2010 ACCF/AHA/AATS/ACR/ASA/SCA/SCAI/SIR/STS/SVM Guidelines for the diagnosis and management of patients with thoracic aortic disease. A report of the American College of Cardiology Foundation/ American Heart Association task force on practice guidelines, American Associations for Thoracic Surgery, American College of Radiology, American Stroke Association Society of Cardiovascular Anesthesiologists, society for cardiovascular angiography and interventions, society of interventional radiology society of thoracic surgeons, and society for vascular medicine. J Am Coll Cardiol 2010;55:e27-129.
17. Ware SM, Peng J, Zhu L, Fernbach S, Colicos S, Casey B, *et al*. Identification and functional analysis of ZIC3 mutations in heterotaxy and related congenital heart defects. Am J Hum Genet 2004;74:93-105.
18. Fulcher AS, Turner MA. Abdominal manisfestations of situs anomalies in adults. Radiographics 2002;22:1439-56.
19. Coral G, Labra A, Schiappacasse G. Abdominal manisfestations of situs ambiguous abnormalities in adults. A study of four cases. Rev Chil Radiol 2013;19:38-43.
20. Kim K, Choi J, Doo Y, Yun YS, Kim J, Lee JB. Well-tolerated and undiscovered common atrium until late adulthood. J Cardiovasc Ultrasound 2016;24:243-6.
21. Zyang Y, Yung ZG, Yang MX, Shi K, Li R, Diao KY, *et al*. Common atrium and the associated malformations: Evaluation by low-dose dual-source computed tomography. Medicine (Baltimore) 2018;97:e12983.
22. Ferdman DJ, Brady D, Rosenzweig EB. Common atrium and pulmonary vascular disease. Pediatr Cardiol 2011;32:595-8.
23. Jiang H, Wang H, Wang Z, Zhu H, Zhang R. Surgical correction of common atrium without noncardiac congenital anomalies. J Card Surg 2013;28:580-6.
24. Hirai S, Hamanaka Y, Mitsui N, Isaka M, Mizukami T. Surgical repair of a common atrium in an adult. Ann Thorac Cardiovasc Surg 2003;9:130-3.
25. Tamura M, Menahem S, Brizard C. Clinical features and management of isolated cleft mitral valve in childhood. J Am Coll Cardiol 2000;35:764-70.
26. Moura-Ferreira S, Sampaio F, Ribeiro J, Fontes-Carvalho R. A rate case series of mitral valve clefts diagnosed by 3D echocardiography and mini-review of the literature. Echocardiography 2019;36:1203-7.
27. Fraisse A, Massih TA, Bonnet D, Sidi D, Kachaner J. Cleft of the mitral valve in patients with Down's syndrome. Cardiol Young 2002;12:27-31.

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