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A Case Report of Left Atrial Isomerism in a Young Patient

Mahmoud Hassan Mahmoud*, Abdullah Majed AlKhudair[1], Fatmah Fareed Almohaisen[2], Reham Abdullah Alshablan[3]

Corresponding Author: Mahmoud Hassan Mahmoud, cardiologist, echocardiography specialist at King Fahad Hospital of University, Eastern Province, Saudi Arabia.

1. Abdullah Majed AlKhudair, a medical intern at King Fahad Hospital of University, Eastern Province, Saudi Arabia.

2. Fatmah Fareed Almohaisen, Echocardiogram technician at King Fahad Hospital of University, Eastern Province, Saudi Arabia.

3. Reham Abdullah Alshablan, Medical student at Imam Abdulrahman bin Faisal University, Eastern Province, Saudi Arabia.

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Abstract

Heterotaxy syndrome (HS) is a rare syndrome and has a huge impact on life. HS refers to the abnormal poisoning of the thoracoabdominal organs. Early detection of HS is crucial and an early intervention and multidisciplinary team follow-up could lead to a better prognosis. HS symptoms vary depending on the type as there are two types, Left Atrial Isomerism (LAI) and Right Atrial Isomerism (RAI). In this report, we present a case of left atrial isomerism in a young patient with multiple surgical corrections along with further HS lifelong complications.

Keywords*: Heterotaxy syndrome (HS), right atrial isomerism (RAI), left atrial isomerism (LAI), and situs inversus.*

Introduction:

Body organs are positioned in specific areas among the normal population. Heterotaxy syndrome (HS) refers to the abnormal position of thoracoabdominal organs. The regular arrangement of body organs is called situs solitus. In HS, the body organs are mispositioned or could be missing **[**1**]**. HS is a rare congenital anomaly that could occur in 1 per 10,000 to 40,000 live births. HS accounts for approximately 3-6% of all congenital heart defects **[**2-4**]**. HS has two main types related to heart malformation, which are right atria isomerism (RAI) and left atria isomerism (LAI). Atria isomerism is defined as having two right or left atria in the heart instead of one right and one left atrium **[**5**]**. In this report, we will focus on the left atrial appendage isomerism in specific. Left atrial isomerism is defined as bilateral left atria in the heart; left atrial isomerism will result in an abnormal venous system, especially in the inferior vena cava. Also, cardiac arrhythmias are more common in patients with left atrial isomerism due to the absence of a sinus node, which is normally located in the right atrium. Non-cardiovascular anomalies present in HS are respiratory, splenic (asplenia or polysplenia), and gastrointestinal anomalies (biliary atresia and malrotated intestines) **[**11**]**.

Case Presentation:

Our case is about a 22-year-old gentleman diagnosed with heterotaxy syndrome as left atria isomerism (polysplenia) complicated by cardiovascular and non-cardiovascular malformations which required surgical intervention early in life. The patient gave a history of hemoptysis in 2020 with an estimated amount of 400ml; the patient was admitted as a case of community-acquired pneumonia. In 2021, the patient had the same complaint, and was managed in OPD as a case of community-acquired pneumonia; no complications were mentioned or noted during the two events. The patient has a history of solitary kidney, and multiple surgical interventions for cardiac anomalies including mitral valve repair 'MVr, aortic valve repair 'AVr, and maze operation for the treatment of atrial fibrillation and subaortic membrane resection. The patient gave a history of renal artery embolism and pulmonary embolism in 2018. In addition, the patient has atrial fibrillation, and he is on warfarin 3mg with a therapeutic INR level. On examination, the patient was in good body shape. Height of the patient: 164cm. Weight: 71kg. BMI: 26.40kg/m2. Blood pressure at the time of the study: 119/66 mmHg. Heart rate: 66 BMP with paced rhythm. Cardiac examination: patient looks normal, not in pain nor respiratory distress, with signs on sternotomy noted on inspection, apex beat palpation, and auscultation. Peripheral examination revealed finger clubbing. Echocardiography was done with the following results:

IVSd	1.2cm	LVd Mass (ASE)	283g	LVSVI (cube)	21.3 ml/ m^2
LVIDd	5.0cm	LVd Mass index ASE	160g/m^2	LVES (cube)	30%
LVIDs	4.5cm	LVESV	90.0ml	LA diameter	4.7cm
LVPWd	1.5cm	LVSV	37.7ml	LVOT diameter	2.4 cm

Table 1. Echocardiography measurements in 2D *Mode.*

LVEDV (4D Auto LVQ)	207.8 ml	LVEDV (4D Auto LVQ)	31%	LVCO (4D Auto LVQ)	3.54 l/min
LVESV (4D Auto LVQ)	143.0 ml	LVESV (4D Auto LVQ)	64.8 ml		

Table 2. Echocardiography measurements in *4D Mode.*

LVOT Vmax	1.24 m/s	LVOT CO	7.56 l/min	TR max PG	47.9 mmHg
LVOT Vmean	0.86 m/s	LVOT Cl	4.27 l/min/ m^2	RAP	3.00 mmHg
LVOT max PG	6.14 mmHg	AV Vmax	1.74 m/s	RVSP	50.99 mmHg
LVOT mean PG	3.38 mm Hg	Dimensionless index	0.71	PV Vmax	1.08 m/s
LVOT VTI	30.5cm	Av max PG	12.07 mmHg	PV max PG	4.69 mmHg
LVOT SV	140.0ml	LV HR	54 bpm	PV Acc. Time	108 ms
LVOT SVI	79.1 ml/ m^2	TR Vmax	3.34 m/s	PV acc. slope	7.51 m/s^2

Table 3. Electrocardiography measurements in Doppler Mode.

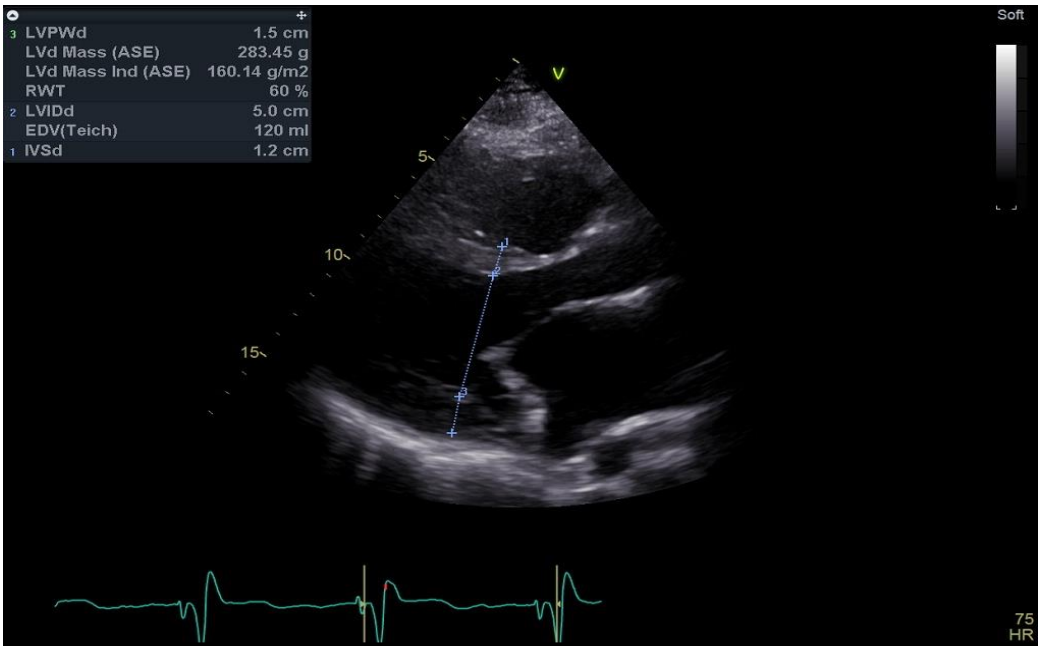


Figure 1: Parasternal long-axis view of left ventricle chamber quantification.

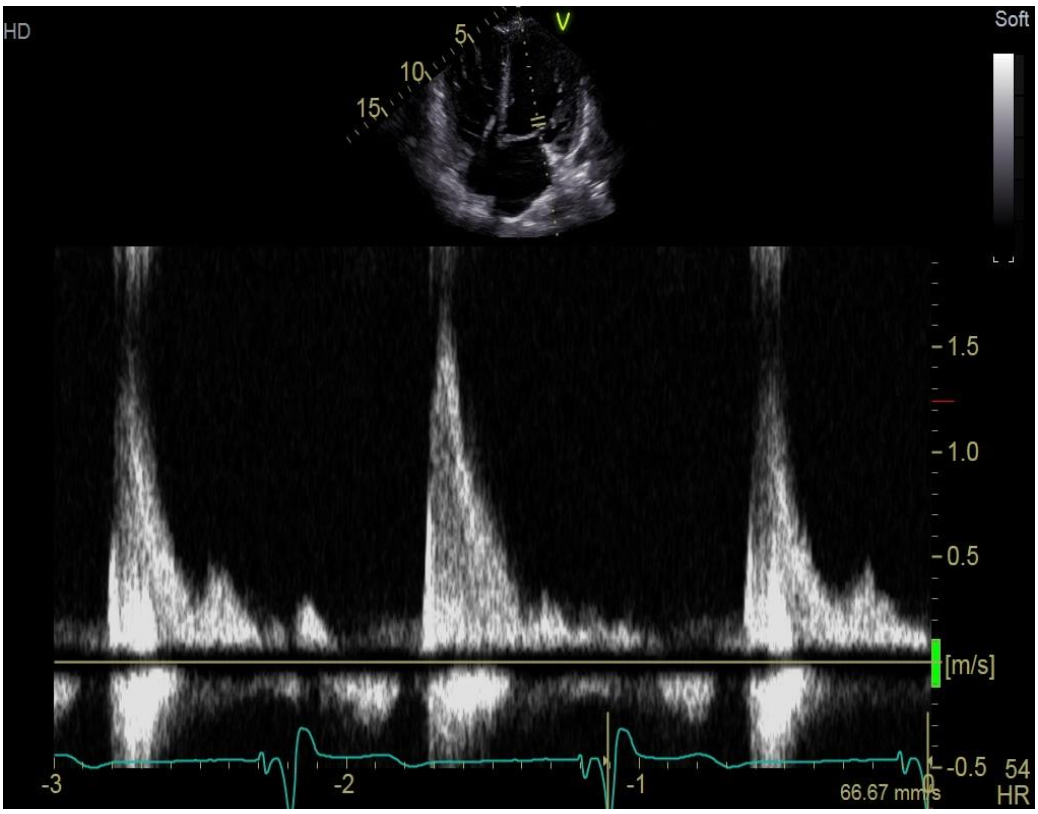


Figure 2: Pulsed wave Doppler of mitral inflow.

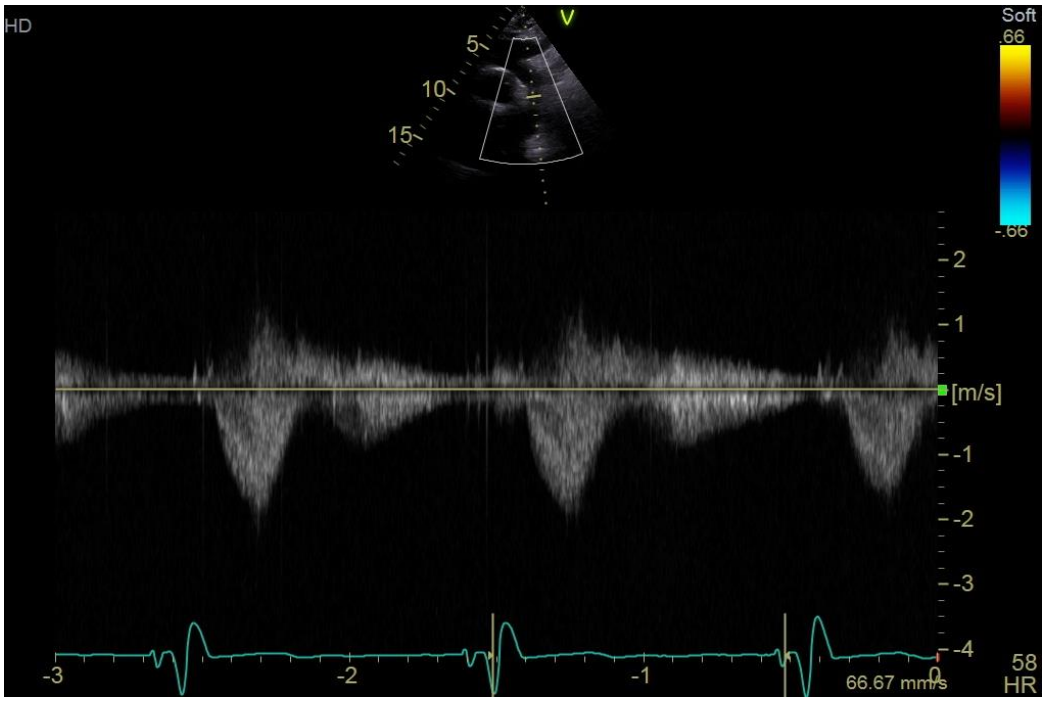


Figure 3: Suprasternal view with continuous-wave doppler on the aorta.

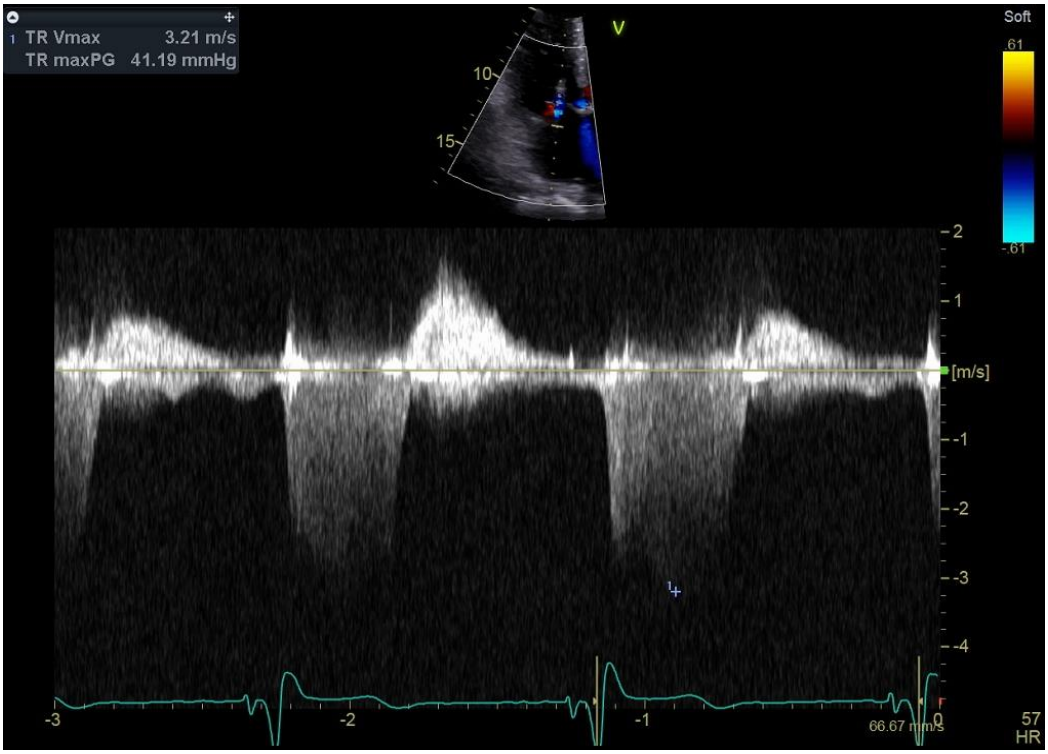
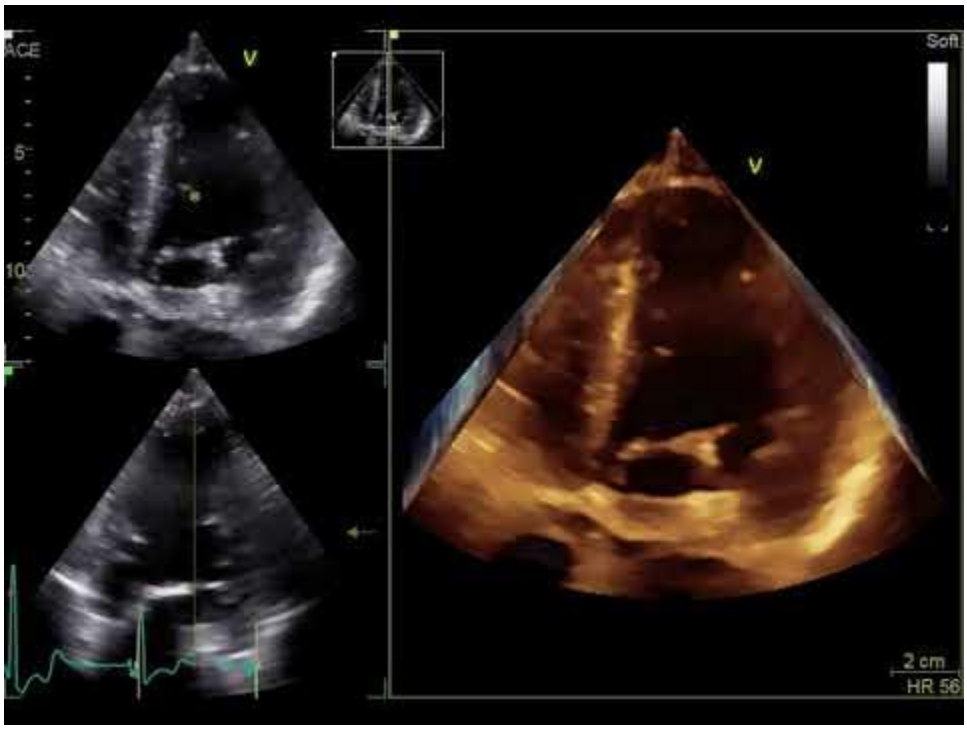
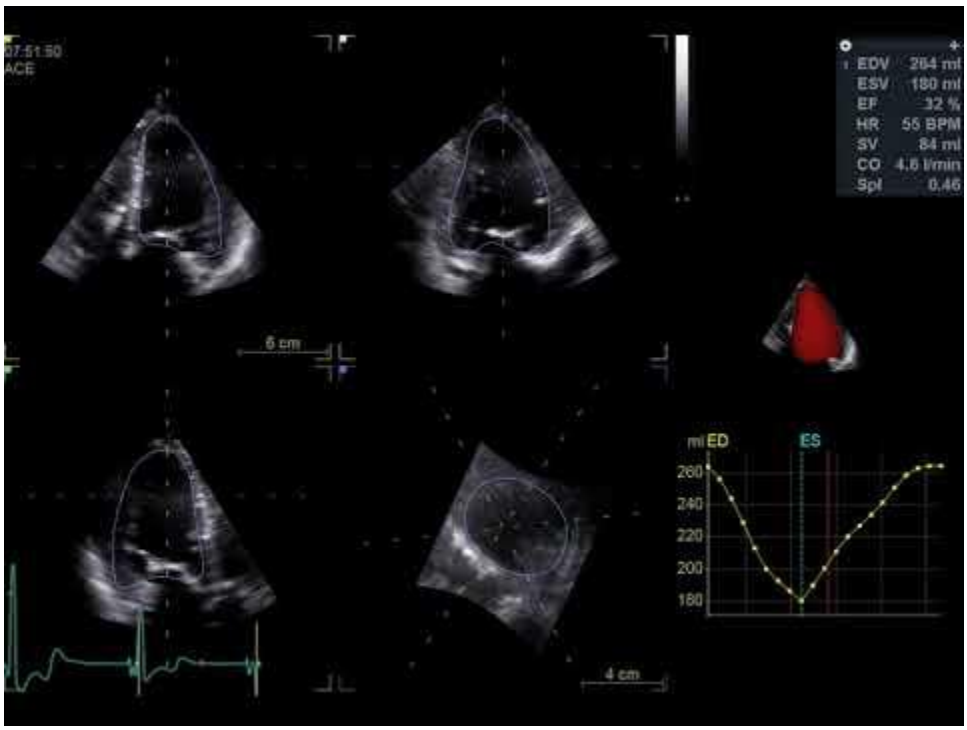


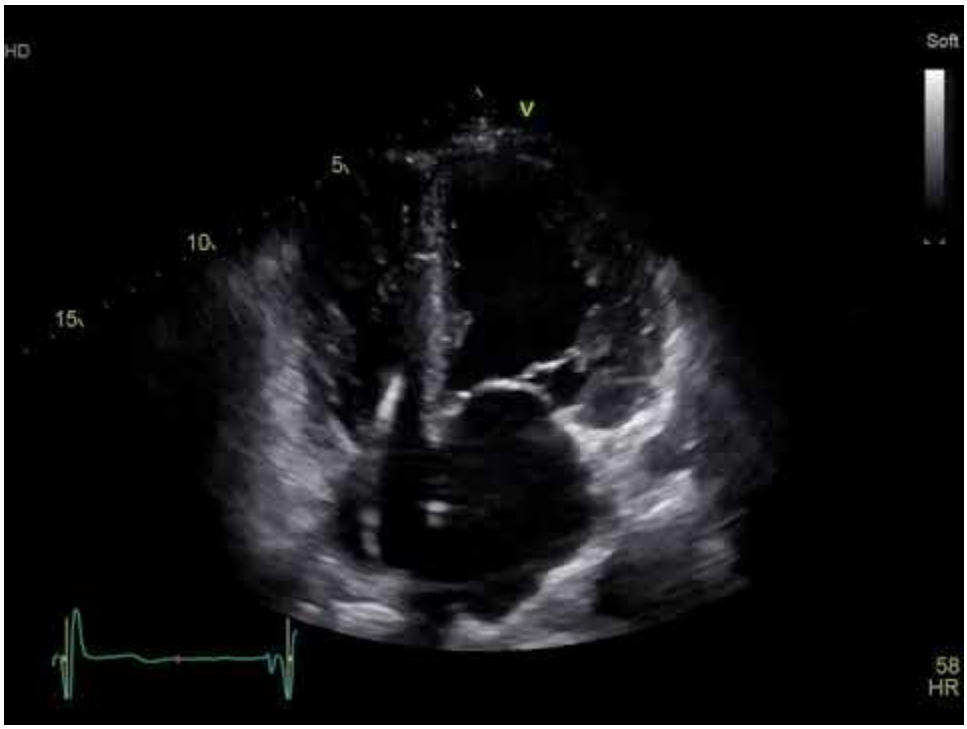
Figure 4: Continuous wave doppler on the tricuspid valve.



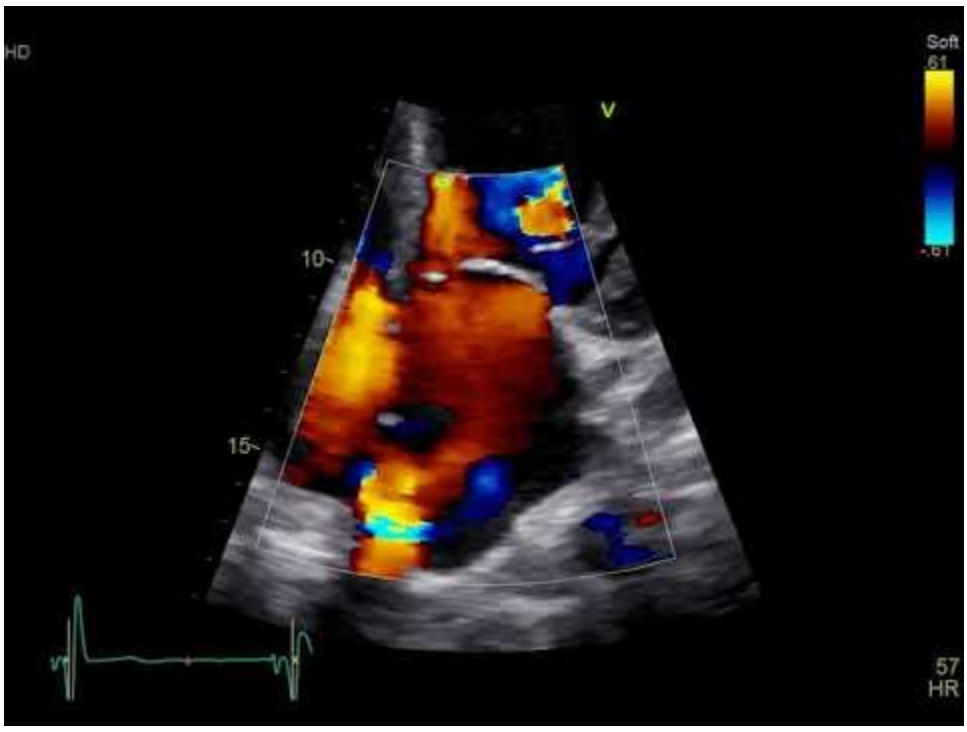
Video.1 3D view of the left ventricle.



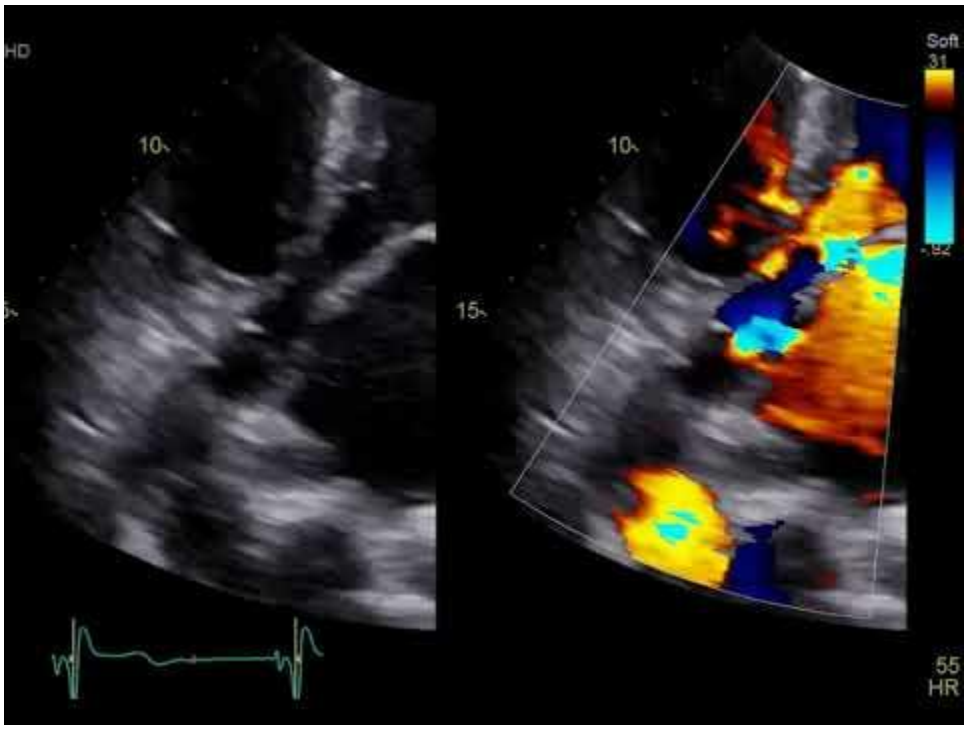
Video.2 4D quantitative of left ventricle ejection fraction.



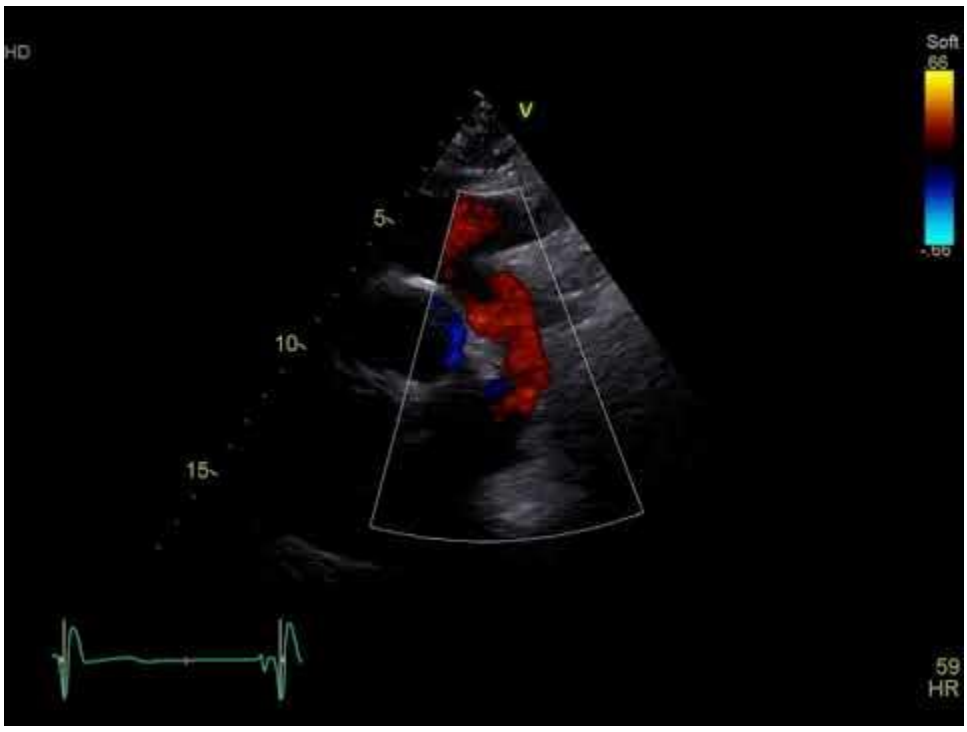
Video.3 Apical 4 chambers view.



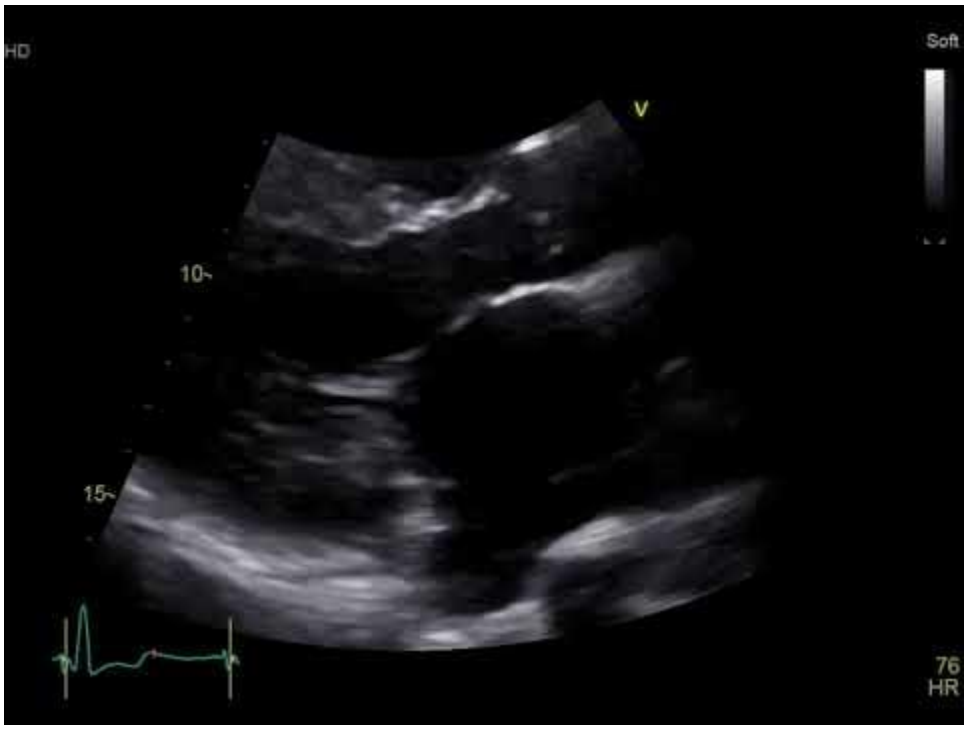
Video.4 Color doppler on mitral valve interatrial septum and tricuspid valve.



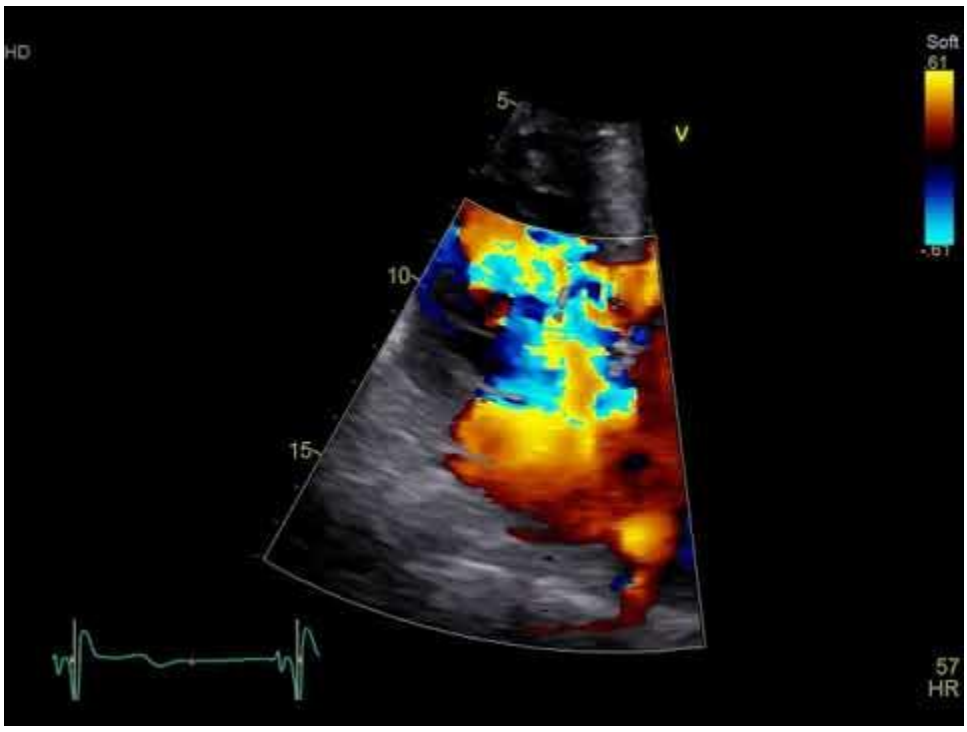
Video.5 Apical 5 chambers view with color doppler on the aortic valve.



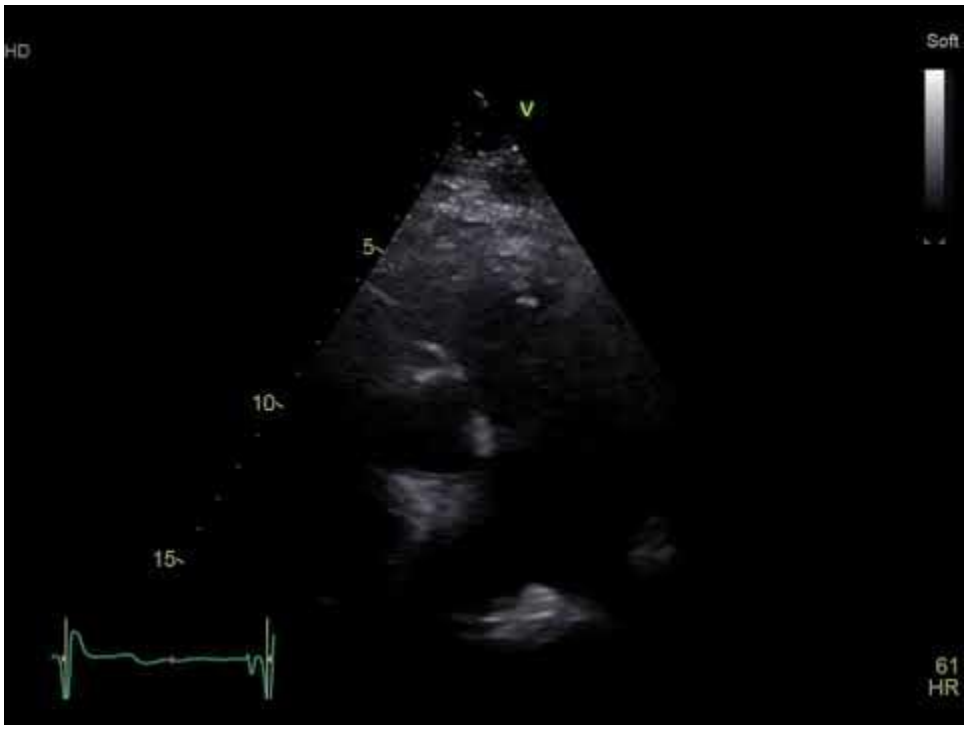
Video.6 Suprasternal view with color.



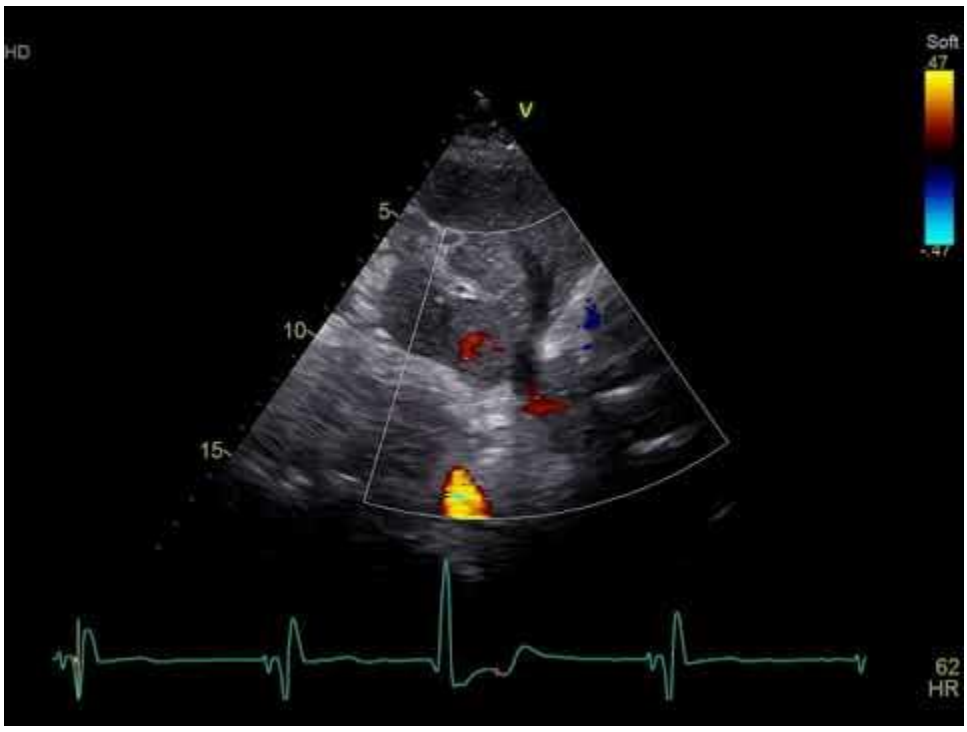
Video.7 Parasternal long-axis view.



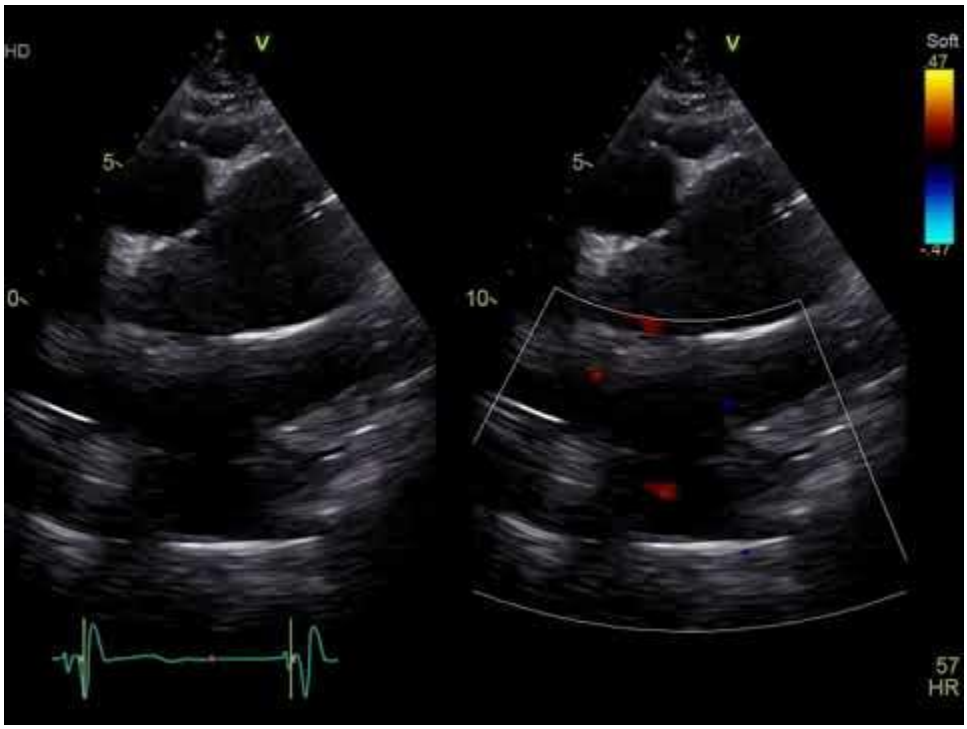
Video.8 Color doppler on the tricuspid valve.



Video.9 Parasternal short axis view with color flow doppler on the pulmonary valve.



Video.10 Subcostal view.



Video.11 Crap view for pulmonary veins drain into the left atrium.

Echocardiography showed a picture of complex congenital heart disease, and the left ventricle was moderately dilated with severe concentric hypertrophy (Figure.1) and it was associated with moderate global hypokinesis (Video.1). Left ventricle ejection fraction was moderately reduced (LVEF: 32%) (Video.2), and regional systolic function assessment was significant for septal motion abnormality consistent with post-cardiac surgery and pacing, with indeterminate diastolic function (Figure.2). The right ventricle was mildly dilated and heavily trabeculated (Video.3), with severely reduced systolic function. The left atrium showed normal cavity volume; the right atrium showed normal cavity volume with pacemaker wire seen in the right atrial cavity (Video.3). A pacemaker wire was also seen in the right ventricle cavity (Video.3). Mitral valve: thickening of the mitral leaflet (Video.3). Post mitral repair with a trivial residual shunt of the cleft anterior mitral leaflet (Video.4). No evidence of mitral stenosis (Video.4). Mild mitral regurgitation was noted (Video.4). The aortic valve showed thickened leaflets (Video.5). No evidence of aortic stenosis. Moderate aortic regurgitation was noted (Video.5). Diastolic flow reversal is present in the descending aorta (Figure.3 - Video.6). Aorta: normal-sized aortic root (Video.7). A tricuspid valve was seen with restricted cusp mobility (Video.3). No evidence of tricuspid valve stenosis. Mild tricuspid regurgitation (Video.8) associated with increased pulmonary pressure RVSP 50.99 mmHg (Figure.4). Thickened pulmonary valve is seen. No evidence of pulmonary valve stenosis. Mild pulmonary valve regurgitation (Video .9). The interatrial septum was seen; it showed a surgically corrected AVSD with a residual shunt (Video.4), and small turbulence at the anterior mitral valve leaflet suggestive of a small cleft mitral valve (Video.4). No signs of pericardial effusion. Systemic veins were interrupted; there

was evidence of double inferior vena cava with the azygous vein (Video.10), and direct drainage of the hepatic vein into the right atrium through a common trunk due to absent hepatic segment of the inferior vena cava (Video.10).

Pulmonary veins showed possibly corrected partial anomalous pulmonary venous return (Video.11).

Discussion:

We are reporting a case of a rare syndrome with various congenital anomalies, including cardiovascular and non-cardiovascular structures. Heterotaxy syndrome is a rare congenital anomaly that occurs in 1 per 10000 to 40000 live births with higher cases of RAI than LAI. Left isomerism, one of the subdivisions of heterotaxy, is associated with paired left-sided viscera and results in bilateral left-sided atrial appendages with no right-sided structures. LAI etiology is still not fully known. However, some studies suggest that LAI might be caused by specific genetic mutations such as ZIC3, NODAL, CFC1, and ACVR2B genetic mutations with different types of inheritance, including autosomal dominant autosomal recessive, and X-linked recessive **[**6,7**]**. Our patient was diagnosed with left atrial isomerism, one of the two types of heterotaxy syndrome (left and right atrial isomerism). Early detection of those patients is crucial for a good prognosis. Antenatal can help in early detection for those who have a family history of HS **[**10**]**. The primary management of heterotaxy syndrome is the surgical correction of the malformed organs (heart, lungs, and others). Medical management and surgical intervention differ according to the severity of the case, with higher rates of operations for heterotaxy with RAI rather than LAI. Prognosis differs from RAI to LAI; a study suggests that RAI has an 85% mortality rate in their first year of life, whereas LAI has a 50% mortality rate. In both conditions, most cases require a total anomalous pulmonary venous connection (TAPVC) repair initial in life and an AV valve repair as it is a long-term risk factor. LAI patients, as discussed earlier, have high rates of experiencing arrhythmias and complete heart block; therefore, they are candidates for a pacemaker insertion as those patients have a higher risk of developing arrhythmias due to the absence of the sinoatrial node **[**8-10**]**. Our patient has had multiple previous surgical procedures, including pacemaker implantation and valvular plastics. Surgical intervention should be augmented with medical treatment to control the manifestations of this syndrome. The pharmacological agents used in our patient are the following: atorvastatin, allopurinol, hydralazine, isosorbide dinitrate, alfacalcidol, levothyroxine, warfarin, and Ventolin. In our patient, we recommend the following: as the patient is a known case of heart failure with reduced ejection fraction (31%), we recommend close follow-up with the cardiology department for frequent assessment of heart function, including the left ventricle function, valvular status, and rhythm assessment. Also, as the patient has a solitary kidney with stage 4 chronic kidney disease, we recommend close follow-up with the nephrology department for proper management of chronic kidney disease and for the consideration of renal transplant therapy. The patient is also a known case of atrial fibrillation and has renal artery

thrombosis, therefore internal medicine/hematology follow-up is essential to maintain a therapeutic coagulation profile and to prevent other venothrmboembolism events. As these patients are at a higher risk of infective endocarditis, we also recommend using prophylactic antibiotics, especially as the patient has a prosthetic repair. Fluid restriction is encouraged to decrease the risk of developing decompensated heart failure. Pneumococcal vaccine and influenza vaccine should be administered in these patients to decrease the risk of pneumonia.

Conclusion:

HS syndrome is a rare congenital anomaly that has a significant impact on life. Early detection and intervention are essential for better outcomes especially surgical correction of the malformed organs and supported by the optimal medical treatment to prevent further complications and maintain a good lifestyle. Antenatal care needs to be closer, and more frequent pregnancy ultrasounds are needed for suspected patients. In addition, searching for other organ anomalies is vital as this syndrome has no standard, well-established clinical features.

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