**CASE REPORT**

*Situs ambiguous* in an Adult with Congenital Cardiovascular and Musculoskeletal Defect

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**ABSTRACT**

*Situs ambiguous* (SA) is an abnormality in which the thoracic and abdominal organs are not clearly lateralized. *Situs ambiguous* with polysplenia is usually associated with complex cardiovascular abnormalities and rarely found among adults due to high mortality at younger age. We are reporting a rare cluster of *Situs ambiguous* with polysplenia (left isomerism) in an adult with congenital hand deformity and cardiovascular pathology, who presented with ascites and decompensated congestive cardiac failure due to dilated cardiomyopathy. He had congenital agenesis of left hand fingers and hypoplastic left thumb. Electrocardiogram and imaging findings were consistent with dextrocardia, *Situs inversus* and features of left isomerism. Echocardiography showed severe biventricular failure with prominent coronary sinus. Such a combination of musculoskeletal and cardiovascular pathologies was not reported in the literature. His coronary arteries were normal. He had successful single chamber Implantable Cardioverter Defibrillator (ICD) insertion.


**INTRODUCTION**

*Situs ambiguous* (SA) is defined as an abnormality in which the thoracic and abdominal organs are not clearly lateralized.1 *Situs ambiguous* with polysplenia is usually associated with complex cardiovascular abnormalities and rarely found among adults due to high mortality at younger age.2 We are reporting a rare cluster of *Situs ambiguous* with polysplenia (left isomerism) in an adult with congenital hand deformity and cardiovascular pathology, who presented with decompensated congestive cardiac failure due to dilated cardiomyopathy. Such a combination of musculoskeletal and cardiovascular pathologies is not reported in the literature at the time of reporting.

**CASE REPORT**

A 32-year man presented to the emergency room with symptoms consistent with decompensated congestive heart failure since 3 - 4 weeks. He had congenital left hand deformity (Figure 1) and had gunshot injury in the chest 20 years back which led to the discovery of dextrocardia (Figure 2).

On examination, he was alert and oriented. He had elevated Jugular Venous Pressure and pedal edema. His blood pressure was 96/70 mmHg; pulse was 90/minute and regular; and $O_2$ saturation was 96% on room air. Chest examination showed bilateral crepitation involving the lower half of the chest, right sided displaced apex beat, which was heaving in nature with a parasternal heave. First and second heart sounds were audible with a third heart sound gallop. Abdominal examination showed distended abdomen with hepatomegaly and ascites. His chest X-ray showed dextrocardia, cardiomegaly and bullet in the left side of chest. Electrocardiographic finding were typically consistent with dextrocardia. Transthoracic Echocardiography (Figure 2) showed dextrocardia, dilated and severe biventricular failure with moderate mitral regurgitation and prominent coronary sinus. Computed Tomography (CT) scan of the abdomen (Figure 3) showed right sided multiple spleens (at least six), right sided stomach and midline liver. CT scan of the chest showed three lobes in

Figure 1: Hypoplastic left thumb and left finger agenesis.
the left lung and two lobes in the right lung, dextrocardia and bilateral superior vena cava. His laboratory investigation showed elevated BNP and liver enzymes. A coronary angiogram showed normal coronaries and venogram showed right superior vena cava opening in left sided morphologic right atrium via coronary sinus while left superior vena cava opening directly to left sided morphologic right atrium.

He was pharmacologically treated for decompensated heart failure with optimum medications. He had successful single chamber Implantable Cardioverter Defibrillator insertion for left ventricular dysfunction and episodes of non-sustained ventricular tachycardia in morphologic left sided right ventricle through right subclavian vein via coronary sinus.

**DISCUSSION**

*Situs* describes the position of the cardiac atria and viscera. *Situs solitus* is the normal position and *Situs inversus* is the mirror image. *Situs inversus* is present in 0.01% of population and was first described by Matthew Baillie. When *Situs* cannot be determined due to unusual position of abdominal and cardiovascular organs it is known as *Situs ambiguous* or heterotaxy. The incidence of *Situs ambiguous* is estimated to be 1 in 40,000 live births. The two primary subtypes of *Situs ambiguous* include right isomerism, or asplenia syndrome and left isomerism, or polysplenia syndrome.

*Situs inversus* and left heterotaxy are mutually exclusive. This patient had dextrocardia, three left and two right lung lobes, right sided stomach, right aortic arch suggestive of *Situs inversus* however, right sided polysplenia (at least 6 spleens) and mid line liver suggests unclear lateralization or *Situs ambiguous*. The discovery of kindreds in which both heterotaxy and *Situs inversus totalis* occur strongly suggests that these are not truly separate diseases. Moreover, because asplenia and polysplenia can occur in the same family, a patient's splenic phenotype should be viewed as merely one phenotypic aspect of an underlying laterality disorder. This patient also had features of both *Situs inversus* and *ambiguous* with left heterotaxy (isomerism) supporting the notion that it is the different spectrum of the same process.

The prognosis of children with *Situs ambiguous* depends upon the severity of associated congenital heart disease. *Situs ambiguous* with polysplenia is usually accompanied with severe congenital cardiovascular defect (Table I): enough to cause fatality among young age and rarely reported among adults. Published cases of *Situs ambiguous* among adults have described its association with congestive cardiac failure and atrial fibrillation, noncompaction syndrome of ventricular myocardium and bicuspid aortic valve with coarctation of aorta.

**Table I:** Spectrum of cardiac abnormalities among fetes with *Situs ambiguous* and polysplenia.

<table>
<thead>
<tr>
<th>Abnormality</th>
<th>Percentage</th>
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<tbody>
<tr>
<td>Complete atrio-ventricular septal defect</td>
<td>68%</td>
</tr>
<tr>
<td>Complete heart block</td>
<td>38%</td>
</tr>
<tr>
<td>Double outlet right ventricle</td>
<td>23%</td>
</tr>
<tr>
<td>Right ventricle outflow tract obstruction</td>
<td>21%</td>
</tr>
<tr>
<td>Total anomalous pulmonary vein drainage</td>
<td>5%</td>
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</tbody>
</table>
This patient also had agenesis of left hand fingers with hypoplastic thumb. Published literature have illustrated wide spectrum of musculoskeletal abnormalities associated with visceroatrial abnormalities.\(^5,6\) This patient’s hand deformity is similar to one of the deformities described in Poland's syndrome but he did not have any chest deformities which constitute an important part of Poland's syndrome.

In patients with heterotaxy hepatic segment of inferior vena cava can be present or absent;\(^6\) coronary Sinus can be normal, absent, or completely unroofed and Superior Vena Cava (SVC) can be normal or bilateral.\(^4,6\) This patient had bilateral superior venacava with normal IVC; however, interrupted IVC was reported to be present in 80% of the patient with Situs ambiguous and polysplenia.

**REFERENCES**


