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HETEROTAXY SYNDROME WITH LEFT ISOMERISM – A CASE REPORT

Radio-Diagnosis

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ABSTRACT

Background: The term "heterotaxy syndrome" comes from the Greek words heteros, which means "different," and taxis, which means "arrangement," and describes a number of thoracoabdominal organ position and morphology abnormalities that do not correspond to the usual lateral positioning order of organs (situs solitus) or its mirror image (situs inversus). This syndrome has two recognised variations: left isomerism, also known as polysplenia syndrome, and right isomerism, also known as asplenia syndrome. Due to less severe cardiac problems in the left isomerism, it has a better prognosis than right isomerism. **Case presentation:** We present a case of a 18-year-old male who came with alleged history of RTA with incidental finding of midline liver occupying right and left hypochondrium and two splenunculi in right hypochondrium. CECT abdomen and plain CT thorax revealed features of heterotaxy syndrome with left isomerism. **Conclusion:** The heterotaxy syndrome, as defined, is frequently linked to intricate cardiovascular abnormalities. The survival of these patients is projected to improve as a result of recent developments in medical therapy, particularly advancements in cardiac surgery skills and procedures taking place globally.

KEYWORDS

Heterotaxy syndrome, Isomerism, Left isomerism, Case report

INTRODUCTION

The term "heterotaxy syndrome" comes from the Greek words heteros, which means "different," and taxis, which means "arrangement," and describes a number of thoracoabdominal organ position and morphology abnormalities that do not correspond to the usual lateral positioning order of organs (situs solitus) or its mirror image (situs inversus) (1-3). It is a rare condition, and cardiac symptoms are primarily responsible for its severity. This syndrome has two recognised variations: left isomerism, also known as polysplenia syndrome, and right isomerism, also known as asplenia syndrome. Due to less severe cardiac problems in the left isomerism, it has a better prognosis than right isomerism (4).

Left Isomerism

When referring to a congenitally deformed heart, left isomerism is described as a subgroup of heterotaxy in which some paired structures on either side of the left-right axis of the body are symmetrical mirror images of one another and exhibit the morphology of the typical left-sided structures. Patients with isomeric left atrial appendages usually have multiple spleens, as well as bilaterally bilobed lungs with lengthy bronchial tubes.

Patients with isomeric left appendages frequently have connections between their pulmonary veins and both atrial chambers, acting as though both atriums were anatomically left atriums.

Right Isomerism

Regarding the congenitally deformed heart, right isomerism is described as a subgroup of heterotaxy in which some paired structures on either side of the left-to-right axis of the body are symmetrical mirror images of one another and exhibit the morphology of the typical right-sided structures. Patients with isomeric right atrial appendages usually have bilaterally trilobed lungs with small bronchi on either side as well as a spleenless condition. However, there are instances where the sidedness of the atrial appendages does not coincide with the sidedness of the lungs or the spleen.

Case Presentation

An 18 year old male came to the emergency department with alleged history of RTA and sustained blunt trauma to abdomen. E-FAST turned to be negative, however incidental finding of midline liver occupying right and left hypochondrium and two well defined iso-echoic splenunculi in right hypochondrium. CT abdomen was advised for further evaluation. Routine blood investigation showed Hemoglobin (Hb) of 13.9 g/dL (Range 13 - 17), Total Leucocyte Count (TLC) of 6700 (Range 40000 - 11000) and platelet count of 2.41 lakhs/cu mm (Range 1.5 – 4.1). Renal function test showed urea of 20 mg/dl (Range 19 - 43) and creatinine of 0.7 (Range 0.66 – 1.25). Barium meal follow through study revealed right sided stomach and small bowel and large bowel are located predominantly on the left and right side of the abdomen respectively (Figure 1). Contrast enhanced computed tomography (CECT) abdomen was performed and plain study was extended to cover the thorax. CECT abdomen and plain CT thorax revealed centrally located liver occupying both hypochondrium, polysplenia in right hypochondrium, stomach on right side and partial malrotation of bowel loops (Figure 2). Other findings include inversion of SMA-SMV axis (Figure 3), dorsal agenesis of pancreas, bilateral bilobed lungs (Figure 4) with the main bronchi coursing inferiorly to the pulmonary arteries i.e, hyparterial bronchi characterizing heterotaxy syndrome with left isomerism.

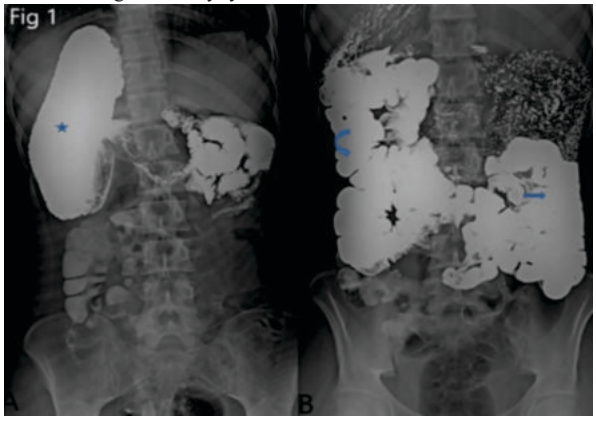

Figure 1 (A, B): Barium meal follow through study. **A** Stomach seen on the right side of midline (blue star) and jejunal loops in the left upper quadrant. **B** Barium meal follow through shows small bowel loops lie predominantly on the left side of the abdomen (straight blue arrow) and large bowel loops lie predominantly on the right side of the abdomen (curved blue arrow).

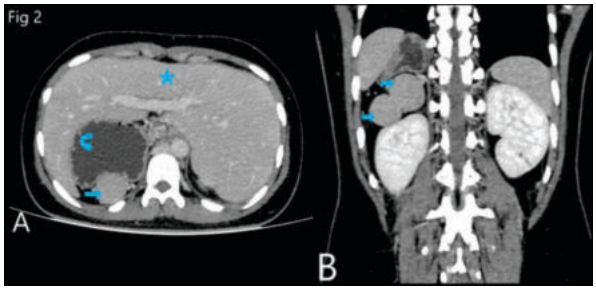

Figure 2 (A, B): Contrast-enhanced computed tomography (CECT) abdomen showing features of heterotaxy syndrome. **A** Axial section of CECT abdomen, portal venous phase showing midline liver (blue star) seen occupying right and left hypochondrium, stomach with hypodense content seen on the right side of midline (curved blue arrow) and splenunculus in the right hypochondrium (straight blue arrow). **B** Post-contrast, coronal reformatted image showing splenunculi (x2) in right upper quadrant (straight blue arrows).

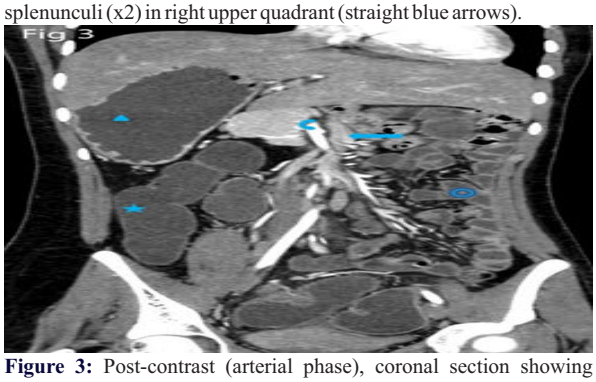

Figure 3: Post-contrast (arterial phase), coronal section showing midline liver seen occupying right and left hypochondrium, stomach with hypodense content seen on the right side of midline (blue triangle), small bowel loops on the left side of midline (blue circle) and large bowel on right side of midline (blue star) with inversion of SMA-SMV relationship i.e, SMA (curved blue arrow) lies on the right and the SMV (straight blue arrow) on the left.

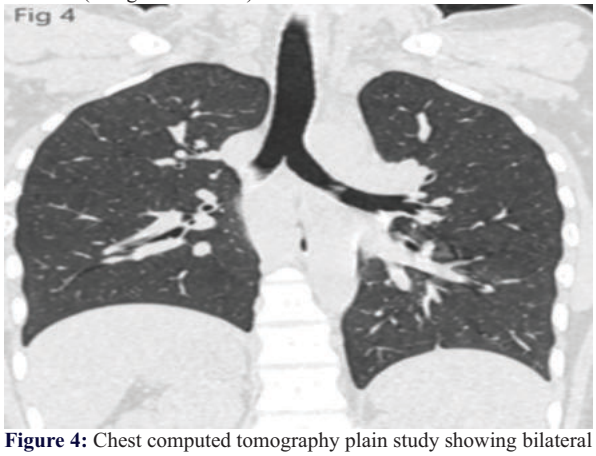

Figure 4: Chest computed tomography plain study showing bilateral bilobed lungs, with main bronchi coursing inferiorly to the pulmonary artery.

DISCUSSION

Incidence And Etiology:

Due to the probable underestimating of the incidence and prevalence, it is challenging to determine the incidence of isomerism with accuracy. Because there are severe forms of cyanotic heart disease present, it appears likely that the majority of right isomerism instances are discovered in infancy. Due to the greater spectrum of related heart problems, including milder kinds that might not even require surgical intervention, it is more likely that left isomerism will go undetected. Contrary to Westerners, Asians exhibit a higher prevalence of heterotaxy syndrome (32%) (5). The emergence of aberrant lateralization and isomerism doesn't appear to have a single etiological cause. Animal and human studies have provided evidence of causal heterogeneity. Visceral heterotaxy is relatively infrequently accompanied by chromosomal abnormalities.

Clinical Features and Imaging Findings

Patients with heterotaxy and complex cardiac abnormalities have a bad prognosis. For patients with asplenia, the 1-year mortality rate is >85%, and for patients with polysplenia, it is >50%. (6). Infants with right isomerism almost invariably have blockage of the pulmonary outflow tract, frequent mixing scenarios, and pulmonary atresia in 25% of instances (7). Despite the fact that cyanosis is by far the most frequent symptom, newborns with right isomerism may exhibit significant respiratory distress as well as cyanosis due to an occluded aberrant pulmonary venous connection. This most frequently occurs when the supracardiac character of the aberrant venous connection. Serious extracardiac abnormalities can occasionally be found in right isomerism patients (8 & 9).

Patients with left isomerism will have non-specific clinical signs that will represent the lesions connected to left isomerism. The cardiac problem might be less severe than right isomerism. In fact, we have seen interrupted inferior caval veins and isomerism of the left atrial appendages but no other cardiac or vascular anomalies. If extracardiac defects, such as biliary atresia or intestinal malrotation, do not call attention to the aberrant positioning of the abdominal organs, such patients may never be diagnosed with isomerism. In a review of all patients diagnosed with isomeric left atrial appendages (10), two-thirds had simpler forms of cardiac disease that were potentially suitable for biventricular repair. The remaining patients had complex cyanotic heart disease, frequently associated with a univentricular atrioventricular connection.

The presentation known as asplenia/right isomerism typically exhibits intestinal malrotation along with trilobed lungs, a left atrium with morphology similar to the right atrium, a liver positioned in the middle, a left-sided aorta, and an inferior vena cava.

In the polysplenia/left isomerism presentation, the liver is also centrally positioned, the right atrium is anatomically identical to the left atrium, the hepatic segment of the inferior vena cava is absent, and there is intestinal malrotation in addition to the duplication of the structures on the left side of the body. Cardiac changes are less common and milder, which accounts for a larger prevalence of such results in people who are older (3).

Treatment:

The kind and severity of the accompanying cardiac and extracardiac abnormalities should be taken into account when deciding how to treat patients with isomerism. Since normal anatomy is rarely achieved, cardiac surgeries for patients with isomerism are typically palliative in character. Thus, it is not unexpected that people with heterotaxy syndrome continue to experience significant death rates. A common atrioventricular valve that is incompetent, abnormalities of the systemic venous connection, a partial or complete anomalous pulmonary venous connection, and a morphological right ventricle supporting the systemic circulation are all factors that have historically been linked to increased operative risk (11 & 12).

CONCLUSION

The heterotaxy syndrome, as defined, is frequently linked to intricate cardiovascular abnormalities. The survival of these patients is projected to improve as a result of recent developments in medical therapy, particularly advancements in cardiac surgery skills and procedures taking place globally. When considered from the moment of birth, additional findings continue to point to an overall bad prognosis for newborns with isomerism, even though surgery statistics point to improved outcomes. Congenital cardiovascular surgeons and paediatric cardiologists continue to face one of their most difficult cases with this condition.

Abbreviations:

E-FAST	Extended Focused Assessment Sonography for Trauma
CT	Computed Tomography
CECT	Contrast enhanced computed tomography
SMA	Superior Mesenteric Artery
SMV	Superior Mesenteric Vein
RTA	Road Traffic Accident

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