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Case Report

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A CASE REPORT ON HETEROTAXY SYNDROME

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ABSTRACT

Heterotaxy syndrome, also known as situs ambiguous, encompasses a group of developmental abnormalities stemming from an aberrant arrangement of thoracoabdominal organs, disrupting their typical position.[1] The etiology is multifactorial, with genetics playing a major role in its presentation.[2] Identification of the syndrome is often incidental when the patient is evaluated for other diseases. The diagnosis of this group of anomalies relies on identifying multiple malformations andmalpositions, often visualized through radiological imaging techniques like ultrasound, computed tomography (CT), or magnetic resonance imaging (MRI). In patients with heterotaxy, the diagnosis and treatment of various commonly acquired illnesses is complicated by abnormal anatomical positioning of the visceral organs. It sometimes leads to delayed or wrong diagnosis, which further hinders timely treatment. Further, the underlying congenital anomalies in various organs complicate the management of even simple conditions. In this case, we look at the multiple challenges we faced as a treating team in accurately diagnosing and treating a patient who presented to the emergency department and was later detected to have heterotaxy.

INTRODUCTION

Heterotaxy refers to a condition where thoraco-abdominal organs deviate from their typical arrangement along the body's left-right axis. The term itself originates from the Greek words "heteros," meaning other than, and "taxis," meaning arrangement.[3] Another term for heterotaxy syndrome is "situs ambiguus," and indicates an intermediate organ arrangement between situs solitus (normal arrangement) and situs inversus (mirror image arrangement of organs).[2]

The normal arrangement of organs in our body is regulated by more than 80 identified genes. A mutation in one or more of these genes can lead to disruption in the pattern of arrangement of organs. Heterotaxy syndromes can occur independently or alongside other genetic conditions like primary ciliary dyskinesia. They may be caused by sporadic genetic mutations or inherited genes, as seen by higher incidence in societies with inbreeding. Some of the commonly identified genes responsible for this syndrome are ZIC3, ACVR2B, LEFTYA (EBAF), CRYPTIC, CRELD1, and NKX2.5.[4]

HS affects various organ systems, resulting in diverse anatomical anomalies with wide-ranging immunological, physiological, and structural implications. The most commonly involved organs are the heart, lungs, liver, kidneys, spleen, gastrointestinal tract, and kidneys, with variations in their structure location, and number.

No single pathognomonic feature clinches the diagnosis of heterotaxy. Rather it is a heterogeneous group with no specific pattern. The most recognized morphological variations that occur can be categorized into asplenia syndrome, polysplenia syndrome, heterotaxy with right atrial appendageisomerism, and heterotaxy with left atrial appendage isomerism, etc.

Diagnosis and management of heterotaxy syndrome demand a comprehensive multi-disciplinary approach due to its complex clinical presentation. The workup entails thorough clinical evaluation, imaging studies, and genetic testing when necessary. Management includes medical therapies to support cardiac and respiratory function and surgical interventions targeting specific organ anomalies that have functional impairment, to improve the overall outcomes and quality of life.

This case report explores the intricacies in the management of a patient with heterotaxy syndrome

complicated by acute acalculous cholecystitis and a large inguinoscrotal hernia. We intend to highlight the diagnostic and therapeutic challenges in this case.

CASE REPORT

A 73-year-old male presented to the Emergency Department with lower chest discomfort, multiple episodes of vomiting, and constipation for the last 3 days. There was no associated fever, jaundice, or abdominal distention. He was a known case of right lower limb amelia, with right hemipelvis hypoplasia, and a large right-sided irreducible congenital inguinoscrotal hernia which has progressively increased in size since birth. There was a history of a similar episode four months ago when he was admitted at another center and was conservatively managed as subacute intestinal obstruction secondary to the large hernia. Physical examination revealed normal vital signs. There was mild periumbilical tenderness on palpation. There was a large right-sided irreducible, non- tender inguinoscrotal hernia with bowel as content.

Blood investigations were within normal limits. The abdominal X-ray showed a few prominent large bowel loops in the hernia sac and right hypochondrium. However, a CT scan of the abdomen revealed several congenital anomalies, including a single left kidney with hydroureteronephrosis, hypoplasia of the right hip bone with complete absence of the acetabulum and right lower limb, as well as malpositioned intra-abdominal organs. The liver was enlarged and displaced to the midline with the gallbladder pushed down and medially to the periumbilical region. There was cholelithiasis with mild pericholecystic fluid and fat stranding, suggestive of acute cholecystitis. The spleen was located in the left lumbar region, with 2 splenunculi. There was also a large defect in the lower abdominal wall with herniation of the sigmoid colon, descending colon, and part of the transverse colon into the left hemiscrotum. Based on the CT findings, the patient was diagnosed with acute calculous cholecystitis, a large irreducible right inguinoscrotal hernia with no features of obstruction or strangulation, and heterotaxy syndrome.

The acute infection was managed conservatively with IV antibiotics and other supportive measures Chest X-ray showed situs solitus. An echocardiogram was taken and cardiac anomalies were ruled out. He responded well to conservative management and was discharged after 5 days in a stable condition with the resolution of symptoms. In view of the complexity of anatomy, a planned operation was deemed beneficial and safe for the patient – hence an interval laparoscopic cholecystectomy was scheduled. He was optimized for the surgery in the meantime.

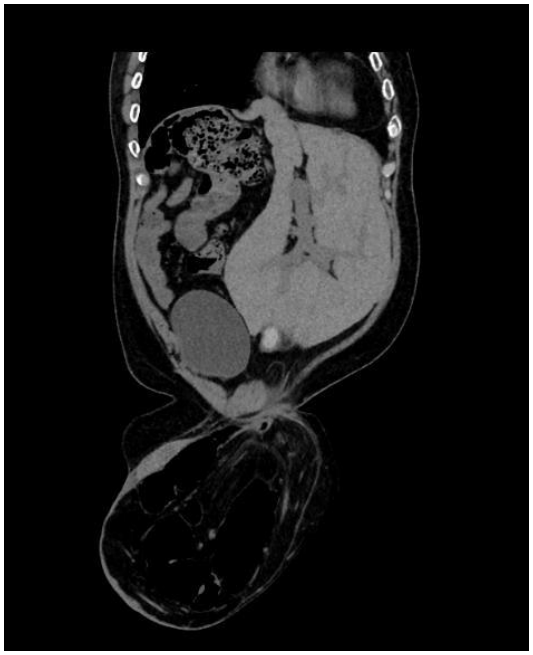

Figure 1: CT Abdomen showing the liver displaced to left and midline with inferiorly located gall bladder showing acute inflammatory changes and stone within. A large right inguinal hernia also seen.

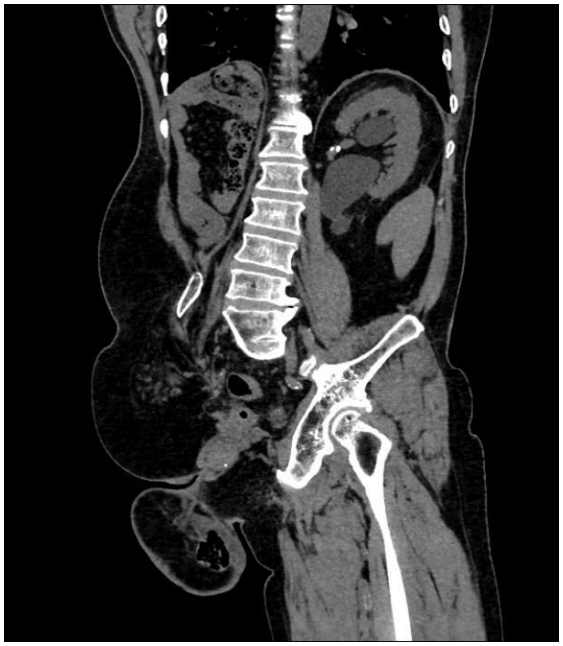

Figure 2: CT Abdomen ashowing single left kidney with moderate hydronephrosis and hypoplasia of right hemipelvis.

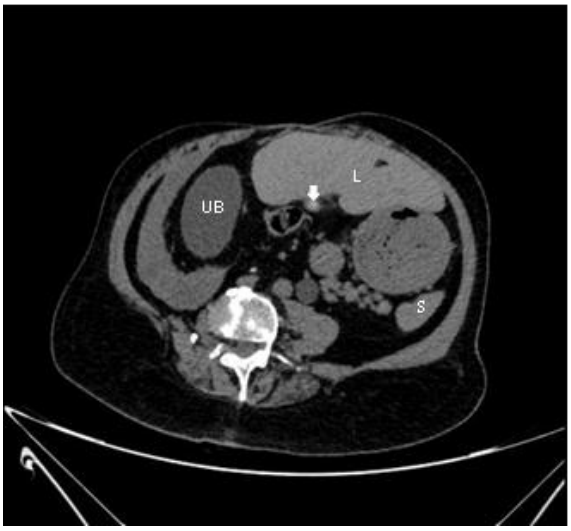

Figure 3: CT Abdomen showingdisplaced displaced spleen (S), liver (L), urinary bladder(UB) and GB(indicated by white arrow) with features of acute calculous cholecystitis.

DISCUSSION

Heterotaxy syndromes pose a multifaceted diagnostic and therapeutic challenge due to its complicated nature, involving the abnormal arrangement of internal thoracoabdominal organs along the body's left-right axis. This condition encompasses various subgroups, such as right isomerism (asplenia syndrome) and left isomerism (polysplenia syndrome), each presenting unique anatomical and clinical characteristics. In adults, these disorders are often incidentally detected while evaluating for some other diseases, as they often have anomalies that are compatible with life, with fewer numbers having significant cardiac and lung anomalies requiring early hospitalization.[5] In our case, the patient's heterotaxy was further complicated by hypoplasia of the right hemipelvis with a large inguinoscrotal hernia, which further contributed to the mal-alignmentof organs.

The clinical symptoms and signs were initially misinterpreted as cardiac or due to intestinal obstruction because of the peri umbilical tenderness in the presence of a large irreducible inguinoscrotal hernia and lower chest discomfort. CT abdomen revealed a midline liver with gallbladder near the periumbilical region with features of inflammation – thus explaining the peri umbilical pain. The CT abdomen was pivotal in clinching the diagnosis and also in ruling out a possibility of obstruction/ strangulation in the large irreducible right inguinoscrotal hernia.

Calculous cholecystitis necessitates early management to prevent complications like gangrene, mucocele, and

rupture progressing into systemic sepsis. Cross-sectional imaging helped us to zeroin on the diagnosis early and act in the nick of time. It also helped us to plan for the operation later. Likewise, the large hernia carried a risk of strangulation and obstruction which would have required emergency surgical intervention. The presence of a single left kidney alerted us to avoid the use of nephrotoxic drugs in the management of infection and pain.

The abnormally placed liver, kidney, pancreas, and spleen with splenunculi, hemi-hypoplasia of the right hip bone, and complete absence of the acetabulum in the right lower limb exemplify the spectrum of anatomical anomalies often associated with heterotaxy syndrome. It points to the need for a comprehensive evaluation of associated abnormalities and a multidisciplinary approach to care.

Addressing the large irreducible right inguinoscrotal hernia and lower abdominal wall defect presents additional challenges. Surgical intervention is crucial but must be approached cautiously given the complex anatomy, size of the hernia, and coexisting congenital anomalies.

CONCLUSION

This case report involves a 73-year-old male patient, who leads a relatively normal life with a myriad of congenital abnormalities.

The presence of heterotaxy syndrome, as evidenced by the patient's CT abdomen findings, highlights the importance of recognizing and understanding the anatomical variations characteristic of this condition. Furthermore, the concurrent diagnosis of cholelithiasis and a large irreducible right inguinoscrotal hernia add layers of complexity to the clinical presentation, requiring careful assessment and prompt intervention to prevent complications.

Through meticulous evaluation, coordinated care, and a multidisciplinary approach, the patient received timely interventions aimed at addressing acute symptoms and optimizing long-term outcomes.

In essence, this case underscores the need for a holistic and multidisciplinary approach to managing complex congenital anomalies such as heterotaxy syndrome. By addressing the unique challenges posed by each patient's condition, it is necessary to improve patient outcomes and enhance overall quality of life.

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