Unraveling Abdominal Heterotaxy Syndrome: A Case Study of Intractable Nausea and Vomiting in an Adult Female

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Background

Heterotaxy syndrome (HS), also called atrial isomerism or situs ambiguus, is a rare congenital condition in which the thoracoabdominal organs and vessels are abnormally arranged across the left-right axis of the body in a non-orderly fashion.[1-5] There is no single phenotype of HS, and clinical severity of the condition is widely variable, dependent on the anatomic derangements present and whether they are producing symptoms. Structural cardiac defects are the main source of morbidity and mortality and are typically identified early on in life in pediatric patients with severe, complete thoracoabdominal heterotaxy. However, another major challenge in management of a broad spectrum of HS patients involves intestinal rotation abnormalities (IRA).[2,5]

IRA are historically managed using a Ladd procedure, named after Dr. William Edward Ladd, who first performed the surgery in the 1930s and involves lysis of adhesions (Ladd's bands), widening of the mesentery, and repositioning of the bowel.[2] Given the cardiopulmonary instability and high post-operative complication risk in patients with complete thoracoabdominal HS, a "watchful waiting" approach is typically applied to asymptomatic patients prior to pursuing surgery. Our case involves an adult female with heterotaxy syndrome isolated to the abdominal organs who presented with recurrent, intractable nausea, and vomiting.

Case Study

Presentation.

A 47F with PMHx GERD, occasional marijuana use, and heterotaxy syndrome of abdominal organs presented to the ED for evaluation of abdominal pain, intractable nausea, and projectile, bilious vomiting for 6 days. This was the patient's second ED visit since onset. She had also been seen 3x in the last 2 years for the same symptoms, treated with mIVF and PRN antiemetics each time before, but only with temporary relief. The patient was following with GI as an outpatient due to her frequently recurring symptoms, and prior outpatient EGD demonstrated "complex duodenal anatomy", including "double duodenal lumen, resulting in duodenal atresia." Prior colonoscopy was unrevealing. Patient was admitted d/t her perceived risk for, as well as concern for, small bowel obstruction.

Objective Findings.

The patient was afebrile, hemodynamically stable, and non-toxic appearing. Her initial work-up was significant for hypertension, hypokalemia, and her abdominal exam was benign. CT A/P showed no e/o bowel obstruction and stable intestinal malrotation, with the small bowel located primarily in the right abdomen and large bowel in the left abdomen. Prior CTs from previous ED visits also discussed findings consistent with heterotaxy syndrome. SBFT confirmed no acute bowel obstruction, with contrast reaching the colon at 2.5 hours. The duodenum was not definitively seen crossing midline in this study, but the duodenojejunal junction was seen positioned to the right of midline in prior CTs.

This research was supported (in whole or in part) by HCA Healthcare and/or an HCA Healthcare affiliated entity. The views expressed in this publication represent those of the author(s) and do not necessarily represent the official views of HCA Healthcare or any of its affiliated entities.

Case Study (continued)

Consultants.

The patient's case was discussed with GI, who determined that her intestinal malrotation (as well as her complex duodenal anatomy) are likely accountable for her recurrent abdominal pain, N/V. Furthermore, IRA also likely causing intermittent vascular obstruction, resulting in episodic ischemia. As a result, surgery was consulted and discussed definitive treatment of her IRA to prevent future symptom recurrence. The patient elected for surgical intervention.

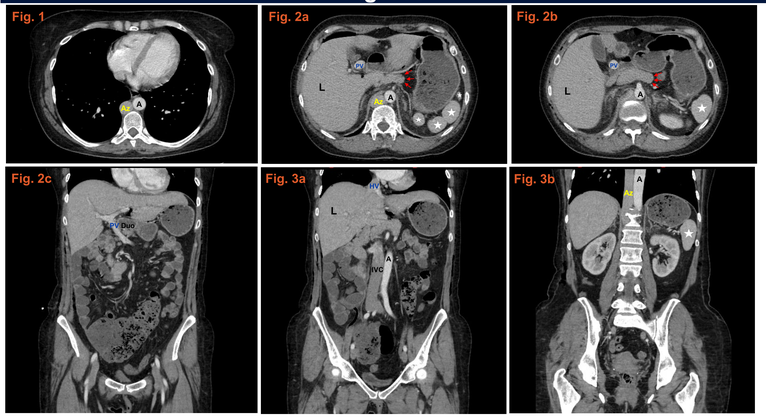
Treatment.

Prior to the OR, patient was managed in the interim with supportive care, including mIVF, IV Zofran, Ativan, and PO Mirtazapine. In the OR, the pt had findings of "bowel malrotation with the colon in the left abdomen, small bowel in the right abdomen, the duodenum adherent to the retroperitoneum, and the Ligament of Treitz on the right side, with sharp angulation, causing the obstruction." The duodenum was freed of its abdominal and retroperitoneal attachments via lysis of Ladd bands.

Post-Operative Course.

Post-operative CT A/P showed no e/o recurrent bowel obstruction. Stable constellation of findings suggestive of heterotaxy, including midgut malrotation, polysplenia, probable dorsal pancreatic agenesis, and IVC interruption with hemiazygos continuation. Pt discharged home on POD#1. Per chart review, pt has not returned with any abdominal pain, nausea, or vomiting since her procedure.

Figures



- **Figure 1**. Axial CT image at the level of the chest, demonstrating normal atrial situs and azygos continuation of the IVC (*Az*), in parallel course with aorta (*A*).
- **Figure 2**. (**a and b**) Consecutive axial CT slices of the upper abdomen showing polysplenia (⋆), truncated appearance of the pancreas (*red arrows*), and preduodenal course of portal vein (*PV*). (**c**) Coronal CT image of the A/P shows preduodenal course of portal vein (*PV*) and bowel malrotation.
- **Figure 3**. (**a and b**) Coronal CT images showing direct continuity of hepatic veins (*HV*) with right atrium in **3a** and azygos continuation of IVC (*Az*) in **3b**.

Discussion

This patient's heterotaxy syndrome was isolated to her abdominal organs, and therefore, she was at no increased anesthetic morbidity or mortality risk for surgery, i.e. she had no structural heart disease. It is important to recognize that heterotaxy is generally classified into two major syndromes: polysplenia and asplenia.[3] Polysplenia is classically termed left isomerism. However, in autopsies of 146 polysplenia patients, only 55% reported bilateral lobed lungs and hyperarterial bronchi; therefore, bilateral left-sidedness is believed to be much less prevalent in adult patients with polysplenia syndrome.[3,6] Moreover, polysplenia is also associated with situs solitus (normal anatomic arrangement), situs inversus (complete left-right reversal), and isolated abdominal heterotaxy.[3] Interestingly, polysplenia does tend to be more common in females, and compared to asplenic patients, cardiac anomalies are less common and usually less complicated in these patients.[3-5] Consequently, echocardiogram may only be clinically indicated in a symptomatic patient with polysplenia syndrome. On the other hand, a clinician might have a lower threshold to obtain an echocardiogram in a patient with congenital asplenia.

Our patient is a prime example of abdominal heterotaxy, and her imaging revealed all findings typically associated with the polysplenia subtype, including, IVC interruption with hemiazygos continuation (**Figs. 1**, **2a**, **3b**), dorsal pancreatic agenesis (**Figs. 2a**,**b**) [the dorsal pancreas and spleen both develop in the dorsal mesogastrium], IRA (**Figs. 2c**, **3a**), and a preduodenal course of the portal vein (**Figs. 2a**,**b**,**c**). IRA are usually collectively termed malrotation but can be further classified to nonrotation (small bowel entirely on the right and colon on the left) or incomplete rotation (bowel appearance between normal and nonrotation).[3] Our patient's anatomy is more consistent with nonrotation. In nonrotation IRA, mesenteric attachment is very narrow and thus predisposes to midgut volvulus, a surgical emergency.[3] Therefore, it is important to radiologically identify intestinal malrotations even in the absence of symptoms due to increased risk and need for surgical correction.

Conclusions

Understanding heterotaxy syndrome and its subdivisions, asplenia versus polysplenia, can help facilitate coordination of care and prompt involvement of surgical specialists in future management of patients with this rare condition. Additionally, polysplenia may be considered a reassuring imaging finding. Lastly, it is of utmost importance for patients to establish care and a strong following with their primary care physician, who can streamline patient work-up and management, including avoiding unnecessary additional radiation exposure from repeated imaging studies.

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