Heterotaxy Syndrome

Incidental Abdominal Heterotaxy Syndrome

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Background

Heterotaxy syndrome, is a sporadic genetic condition with a prevalence of 1:10,000\(^1\). It is characterised by an abnormal arrangement of thoracic or abdominal viscera and can occur in isolation or as a feature of other genetic conditions\(^1\). If any step of embryonic visceral organ arrangement is impaired, abnormal organ position and heterotaxy syndrome may ensue\(^2\).

Findings include structural cardiac defects, lung lobe abnormalities, abnormal spleen development, intestinal malrotation or midline liver position\(^1\). Heterotaxy syndrome should be differentiated from partial malrotations or situs inversus, where there is complete inversion of thoraco-abdominal organs\(^3\).

Case Presentation

NC is a 53F referred by her general practitioner to a general surgery outpatient clinic for an opinion on long-standing, intermittent left sided para-umbilical abdominal pain, described as burning in nature. The pain occurred nocturnally and was aggravated by spicy foods and movement. Panadeine forte provided symptom relief. She denied changes to bowel habit,
Heterotaxy Syndrome

constitutional symptoms, and had no significant family history. Past medical history comprised a hysterectomy eight years prior.

On examination she was vitally stable with mild left para-umbilical tenderness without any palpable masses or guarding. Blood tests revealed a mild anaemia and slightly elevated inflammatory markers. Given her non-specific symptoms she underwent a gastroscopy, colonoscopy and an abdominal computed tomography (CT) scan.

**Outcome**

NC’s gastroscopy and colonoscopy were unremarkable bar some mild gastritis. Gastroscopy was conducted with the patient in the left lateral decubitus position. Conventional upper gastrointestinal endoscopic techniques and movements were difficult to utilise, secondary to mirrored position of relevant viscera. Slight clockwise rotation and upwards deflection (conventionally an anticlockwise rotation and upwards deflection) of the endoscope tip was required to advance from the oesophagus, gastro-oesophageal junction and then into the stomach. This was likely due to the rightward slant of the oesophagus to the right-sided stomach (Figure 1). Furthermore, passage from the duodenal bulb to the descending duodenum was difficult requiring manipulation of the scope tip including counter-clockwise rotation (conventionally a clockwise rotation) and upwards deflection to advance. J- and U-turn manoeuvres to fully appreciate the greater curvature, cardia and fundus needed to be conducted in a mirrored manner using the unique fold arrangement of the greater and lesser
Heterotaxy Syndrome

curvatures as guidance landmarks. Colonoscopy proceeded without issue. Salient findings from her abdominal CT scan comprised an equivocal small fat containing umbilical hernia, right-sided stomach (Figure 1), right-sided spleen situated postero-inferior to the liver (Figure 2), a midline liver (Figure 3), a small spleneculus (Figure 2) and left-sided inferior vena cava (Figure 1, 3). NC was managed with proton-pump inhibitors and had ongoing follow-up to evaluate adverse complications from her anatomical findings.

Based on aforementioned investigations, a diagnosis of gastritis with an underlying congenital heterotaxy syndrome was made.

Discussion

Heterotaxy syndrome arises from failure of intra-abdominal and intra-thoracic structures to rotate appropriately during embryologic development. This may occur due to a series of mutations in genes regulating development of normal left-right axis determination\(^4\). In children, this may correlate with a poor prognosis due to congenital heart disease\(^5\). However, some case reports suggest that these cardiac anomalies bear minimal symptomatic or medical consequence to allow survival into adulthood\(^6\). Other complications include cyanosis, respiratory impairment, increased infection risk, digestive issues or co-morbid neoplastic disease\(^1,7\).
Heterotaxy Syndrome

Patients with heterotaxy syndrome have been historically categorised into right or left isomerism. Right isomerism (asplenia) is characterised by congenital spleen absence and duplication of right sided organs while left isomerism (polysplenia) is typified by multiple accessory spleens and duplication of left sided organs\(^3,8\). Polysplenia is more common in females and has a wider range of clinical manifestations and prognosis compared to their asplenic counterparts\(^9\). Up to 75% of patients with polysplenia have cardiac defects such as endocardial cushion defects or double outlet right ventricle\(^8\).

Based on pre-procedural imaging clinicians may predict difficult endoscopic procedures and pre-empt unconventional movements required to navigate these patient’s variant anatomy. With respect to the current case, images from the CT scan indicate that it may be reasonable to expect that the altered position of stomach, liver and spleen may shift the position of the upper gastrointestinal tract thus making for a challenging gastroscopy. Assumedly, colonoscopy proceeded without difficulty due to the radiographically normal position of rectum and length of colon to the terminal ileum.

**Take Home Message**

Heterotaxy is a rare, phenotypically variable genetic condition with outcomes dependent on associated cardiac and/or extra cardiac anomalies\(^10\). Anatomic variations may lead to
Heterotaxy Syndrome

diagnostic ambiguity and persons with this condition should be analysed closely, particularly for procedural planning prior to invasive medical or surgical interventions.5

References


Heterotaxy Syndrome


Heterotaxy Syndrome

Figure Captions

**Figure 1:** Coronal CT slice of intra-abdominal and intrathoracic viscera. Note the anomalous position of the stomach (star) with a significant diagonally slanted lower 1/3\textsuperscript{rd} oesophageal lumen, and left inferior vena (arrow).

**Figure 2:** Coronal CT slice of intra-abdominal and intrathoracic viscera. Note the spleneculus/spleen (arrows) noted to be posterior and inferior to the liver.

**Figure 3:** Transverse CT slice of intra-abdominal viscera. Note the anomalous position of stomach (star), left sided inferior vena cava (circled), spleneculus (arrows) and midline liver (L).
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