Heterotaxy Syndrome
Incidental Abdominal Heterotaxy Syndrome

Janaka Lovell¹, Jonathan Sivakumar², Shiran Wijeratne³

¹ BBiomed, MD, PGrad Dip Surg Anat. Werribee Mercy Hospital, Werribee, Victoria 3030, Australia.

² MBBS, PGrad Dip Surg Anat., MS. Werribee Mercy Hospital, Werribee, Victoria 3030, Australia.

³ MBBS, MRCS, FRACS, FICS, FCSSL. Werribee Mercy Hospital, Werribee, Victoria 3030, Australia.

Corresponding Author

Dr Janaka J Lovell

Department of Surgery

300-310 Princess Highway, Werribee

Victoria, Australia, 3030

Telephone number: +61 448889438

Email address: janakalovell@gmail.com

Words: 750

References: 10

Tables/Figures: 3

Key Words: Heterotaxy Syndrome, Situs Ambiguous, Computed Tomography

This is the author manuscript accepted for publication and has undergone full peer review but has not been through the copyediting, typesetting, pagination and proofreading process, which may lead to differences between this version and the Version of Record. Please cite this article as doi: [10.1111/ans.15406](https://doi.org/10.1111/ans.15406)

Conflict of Interest: None to declare

Acknowledgements: Nil

Background

Heterotaxy syndrome, is a sporadic genetic condition with a prevalence of 1:10,000¹. It is characterised by an abnormal arrangement of thoracic or abdominal viscera and can occur in isolation or as a feature of other genetic conditions¹. If any step of embryonic visceral organ arrangement is impaired, abnormal organ position and heterotaxy syndrome may ensue².

Findings include structural cardiac defects, lung lobe abnormalities, abnormal spleen development, intestinal malrotation or midline liver position¹. Heterotaxy syndrome should be differentiated from partial malrotations or situs inversus, where there is complete inversion of thoraco-abdominal organs³.

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,

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

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 determination⁴. In children, this may correlate with a poor prognosis due to congenital heart disease⁵. However, some case reports suggest that these cardiac anomalies bear minimal symptomatic or medical consequence to allow survival into adulthood⁶. 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 counterparts⁹. Up to 75% of patients with polysplenia have cardiac defects such as endocardial cushion defects or double outlet right ventricle⁸.

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 anomalies¹⁰. Anatomic variations may lead to

diagnostic ambiguity and persons with this condition should be analysed closely, particularly for procedural planning prior to invasive medical or surgical interventions⁵.

References

1. National Institute of Health. Genetics home reference: Heterotaxy syndrome. [Web page on internet]. United States of America. [Updated October 2, 2018; Cited October 2, 2018]. Available from: <https://ghr.nlm.nih.gov/condition/heterotaxy-syndrome#diagnosis>

2. Shiraishi I & Ichikawa H. Human heterotaxy syndrome - from molecular genetics to clinical features, management, and prognosis. *Circ J*. 2012;76:2066–2075.
3. Sutherland MJ & Ware SM. Disorders of left-right asymmetry: heterotaxy and situs invertus. *Am J Med Genet Part C Semin Med Genet*. 2009;151C:307-17.
4. Li Y, Yagi H, Onuoha EO, et al. DNAH6 and Its Interactions with PCD Genes in Heterotaxy and Primary Ciliary Dyskinesia. *PLoS Genet*. 2016;12:e1005821.
5. Kim JY, Youn HJ, Kim GH et al. Incidentally detected situs ambiguous in adults. *J Cardiovasc Ultrasound*. 2011; 19(4):211-215.
6. Moss AJ, Adams FH, Emmanouilides GC, et al. Moss' heart disease in infants, children and adolescents. (4th ed.) 1989. Williams & Wilkins, Baltimore, MD.
7. Lee JM, Lee HS, Kim CD. Situs ambiguous with left sided isomerism. *Digestive and Liver Disease*. 2014;46(7):658.
8. Ghosh S, Yarmish G, Godelman A, et al. Anomalies of viscerotrial situs. *AJR Am J Roentgenol*. 2009;193:1107–1117

9. Applegate KR, Goske MJ, Pierce G et al. Situs revisited: Imaging of the heterotaxy syndrome. *Radiographics*. 1999;19(4).
10. Patel A, Costello JM, Backer CL, et al. Prevalence of noncardiac and genetic abnormalities in neonates undergoing cardiac operations: analysis of the society of thoracic surgeons congenital heart surgery database. *Ann Thorac Surg*. 2016;102:1607-14.

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 1/3rd 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).

ANS_15406_Figure_1.png

ANS_15406_Figure_2.png

ANS_15406_Figure_3.png

Minerva Access is the Institutional Repository of The University of Melbourne

Author/s:

Lovell, J;Sivakumar, J;Wijeratne, S

Title:

Incidental abdominal heterotaxy syndrome.

Date:

2020-05

Citation:

Lovell, J., Sivakumar, J. & Wijeratne, S. (2020). Incidental abdominal heterotaxy syndrome.. ANZ J Surg, 90 (5), pp.911-912. <https://doi.org/10.1111/ans.15406>.

Persistent Link:

<http://hdl.handle.net/11343/286374>