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CASE REPORT Open Access

Heterotaxy polysplenia syndrome presenting with bleeding from the greater curvature of stomach: a case report

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Abstract

Background: The "heterotaxy syndrome" also called "situs ambiguus" is a rare condition in which the internal organs of the chest and abdomen are abnormally arranged. This syndrome is usually associated with a spectrum of health-related conditions involving birth defects in organs like the heart, spleen, liver, lungs and others.

Case presentation: We present a case of a 29-year-old male who came with an episode of massive upper gastrointestinal bleeding. On arrival he was vitally unstable and after resuscitation underwent a contrast-enhanced CT scan examination. The CT scan identified the features of heterotaxy syndrome with left isomerism and an abnormal blush from the branch of splenic artery supplying the greater curvature of the stomach. Embolization of the splenic artery branches was performed with the help of gel foam. No abnormal blush was identified post-embolization. Later on, in his hospital stay the patient went into pulse less electrical activity and died.

Conclusion: Upper gastrointestinal bleed in the patients with heterotaxy syndrome is a rare presentation of a rare condition. Our case highlights the importance of diagnostic and interventional radiology in diagnosing such patients and identifying the site of bleed especially where endoscopic visualization is not possible and managing the bleeding efficiently.

Keywords: Heterotaxy syndrome, Upper gastrointestinal bleed, Greater curvature, Stomach, Case report

Background

"Heterotaxy syndrome" also known as "situs ambiguus" is derived from two Greek words; heteros meaning other and taxis which means arrangement. It is a rare condition in which the internal organs of chest and abnormal are abnormally arranged and is associated with a spectrum of health-related conditions including birth defects in organs like heart, lung, spleen and others [1]. It has a worldwide prevalence of approximately 1 in 10,000 people. The presentation depends upon the involvement of the particular organ; for example, the patients with heart defects present with cyanosis and the lung involvement usually manifests as difficulty in breathing and pulmonary infections [2].

There are two known variants of this syndrome; left isomerism, i.e. polysplenia syndrome and right isomerism, i.e. asplenia syndrome. The prognosis of left is better than right isomerism due to less severe cardiac abnormalities in the former [3]. Rahmani and team reported a very rare case of a young non-cirrhotic woman with left isomerism and presented with a massive upper gastrointestinal bleed due to extensive gastroesophageal varices. Her magnetic resonance venography (MRV) showed situs inversus, polysplenia, interrupted Inferior vena cava (IVC) and extensive vascular abnormalities like absent portal vein and extrahepatic portosystemic shunts [4]. Donnelly and associates also reported a case of a 95-year-old woman who presented with a massive upper

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gastrointestinal bleed and diffusely tender on abdominal examination. Her CT scan of abdomen and pelvis did not show the cause of her bleed but revealed features of left isomerism, i.e. multiple splenic nodules, malrotation of midgut and azygous continuation of IVC [5].

We are reporting the first case of left isomerism variant of heterotaxy syndrome presenting with upper gastrointestinal bleed (non-variceal) from the greater curvature of stomach which was managed successfully with radiological intervention.

Case presentation

A 29-year-old male presented with a history of one episode of massive hematemesis. He was referred from another hospital where his esophagogastroduodenoscopy (EGD) was performed which revealed a large blood clot in the stomach. Due to poor visibility on endoscopic examination, the site of bleed was not identified. On arrival the patient was in shock with a blood pressure of 80/45 mmHg; heart rate was 121 beats per minute. He was short of breath and his respiratory rate was 29 breaths per minute with O_2 saturations at room air of 88%. He was resuscitated with intravenous hydration and vasopressors were started. He was kept nil per oral (NPO); intravenous proton pump inhibitor infusion and broad-spectrum antibiotics were given.

His laboratory data showed haemoglobin levels of 9.8 g/dl (normal range: 12.3–16.6) that dropped from his baseline of 16.1 g/dl. He was transfused one packed cell volume. His plasma lactic acid was reported to be 14.7 mmol/L (normal range: 0.5–2.2 mmol/L). He had one episode of massive hematemesis in the emergency room that lead to endotracheal intubation of the patient to maintain his airway. His chest x-ray (CXR) revealed bilateral lung infiltrates. Blood, urine and tracheal cultures were sent.

A contrast-enhanced computerized tomography scan (CECT) of the abdomen was done to identify the site of bleed which showed features consistent with heterotaxy syndrome, i.e. presence of a midline/transverse liver, polysplenia and hemiazygos/azygos continuation of the inferior vena cava, Fig. 1.

The CECT also showed a significantly distended stomach with a large hematoma and arterial blush along the greater curvature representing active bleed, Fig. 2.

Interventional radiology (IR) team was taken on board and they performed mesenteric angiography which revealed an abnormal blush from a branch of the splenic artery, corresponding with CT blush; this was then embolized successfully using gel foam. Post-embolization showed no further filling of the vessels, Fig. 3.

Post-embolization, there was no further episode of any bleeding. His tracheal cultures showed a growth of Raoultella terrigena which was sensitive to fosfomycin and colistin. His echocardiography showed complete, unbalanced, atrioventricular (AV) canal defect, single atrium, suspicion of total anomalous pulmonary veins, common AV valve, moderate to large size inlet ventricular septal defect (VSD) with bidirectional shunt and systemic right ventricular and pulmonary artery pressures.

He was kept under observation in ICU with the ongoing management for gastrointestinal bleed with proton pump inhibitor infusion. His chest infection was treated with culture-sensitive antibiotics; and for shock, intravenous fluids and vasopressors were continued. Later on, in his ICU stay he went into pulseless electrical activity. His cardiopulmonary resuscitation was performed but the return of spontaneous circulation (ROSC) was not achieved and death was declared.

Discussion

Based on certain morphological features, the patients of heterotaxy syndrome are divided in two sub-types, i.e. bilateral right handedness (right isomerism) and bilateral left handedness (left isomerism). Right isomerism is associated with extensive cardiac malformations as compared to left isomerism [6]. The 1-year mortality in patients of heterotaxy syndrome who have complex heart abnormalities is approximately 85% with right isomerism and more than 50% with left isomerism [7].

Left isomerism as in this case is mostly associated with atrioventricular septal defects (ASVD), interrupted IVC (hepatic part), persistence of left SVC, polysplenia, midline liver, extrahepatic portal vein atresia [8]. The prevalence of polysplenia is 2.5 in 100,000 of live births and is found in approximately 55% of patients with left isomerism [9]. Only less than 1% of the population has abnormalities in IVC and interrupted IVC is one of the most consistent findings after polysplenia [10].

We found only two cases of upper gastrointestinal bleed in our literature search, one due to extensive gastroesophageal varices and the cause of other remained unknown. However, no previous case report identified which shows bleeding from the branch of splenic artery supplying the greater curvature of the stomach along with the other morphological features of polysplenia syndrome making our case one of the rare presentations of heterotaxy syndrome. The knowledge of such case presentations will help the clinicians with prompt identification and appropriate management of patients as highlighted in our case.

Conclusion

Upper gastrointestinal bleed in the patients with heterotaxy syndrome is a rare presentation of a rare condition. Our case highlights the importance of

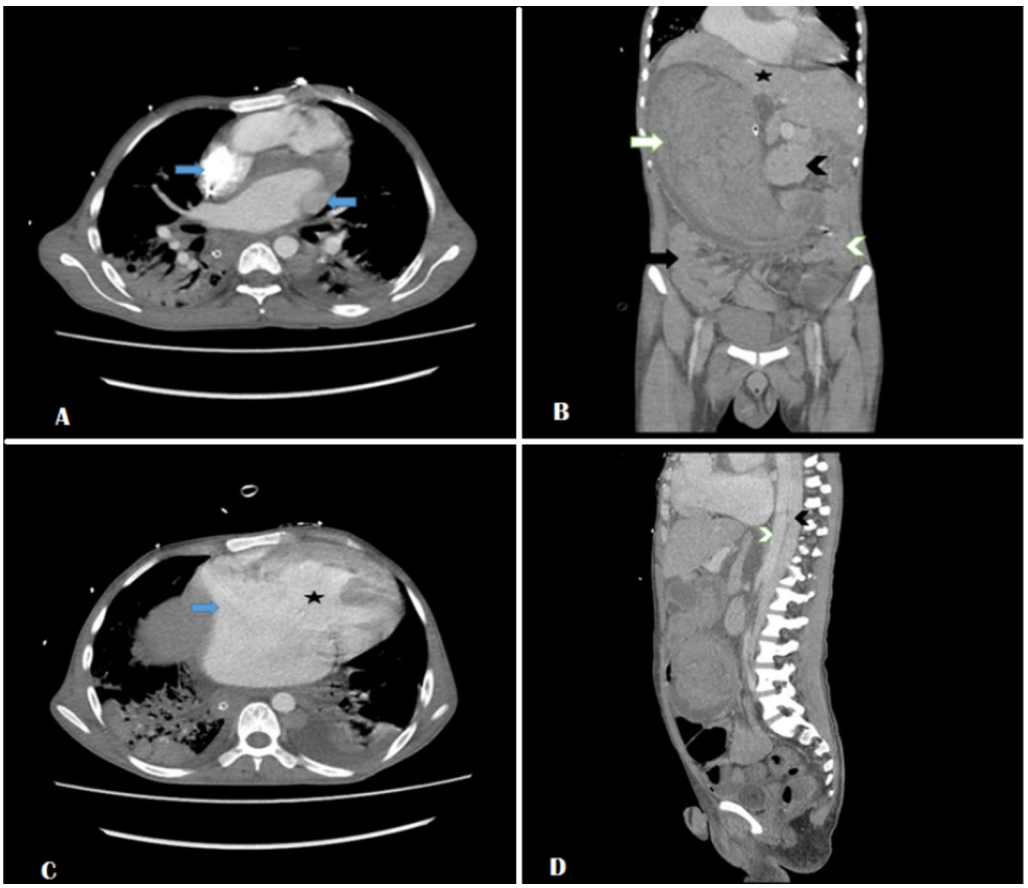

Fig. 1 Contrast-enhanced CT (CECT) showing features of heterotaxy syndrome. **A** Axial section of CECT showing double superior vena cava (SVC) (blue arrows). **B** Post-contrast, coronal section showing midline liver (black asterisk), stomach though distended with high-density content but seen on right side of midline (white arrow), head of pancreas on the left side of midline (black arrowhead), small bowel loops on the left side of midline (black arrow) and large bowel on right side of midline (white arrowhead). **C** Axial section showing large subvalvular ventricular septal defect (VSD) (black asterisk) and dilated right atrium. **D** Post-contrast Sagittal section showing anteriorly placed aorta (white arrowhead) and dilated azygous vein (black arrowhead). Azygous continuation of inferior vena cava can be seen below the level of the diaphragm

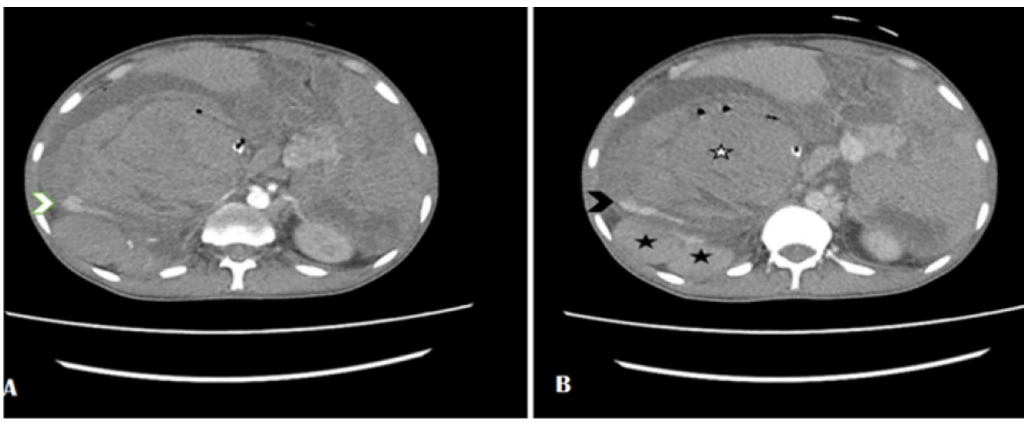

Fig. 2 Contrast-enhanced CT (CECT) to identify any area of active bleeding. **A** Axial section of CECT arterial phase, showing a liner hyperdensity along gastro-splenic ligament (white arrowhead). **B** Axial section from CECT portal venous phase showing increasing linear hyperdensity along gastro-splenic ligament (black arrowhead) representing active bleeding. The stomach is significantly distended with high-density content representing blood clots (white asterisk). Two small-sized splenules are seen (black asterisks) instead of one parent spleen representing polysplenia which is an additional feature of heterotaxy syndrome with left isomerism

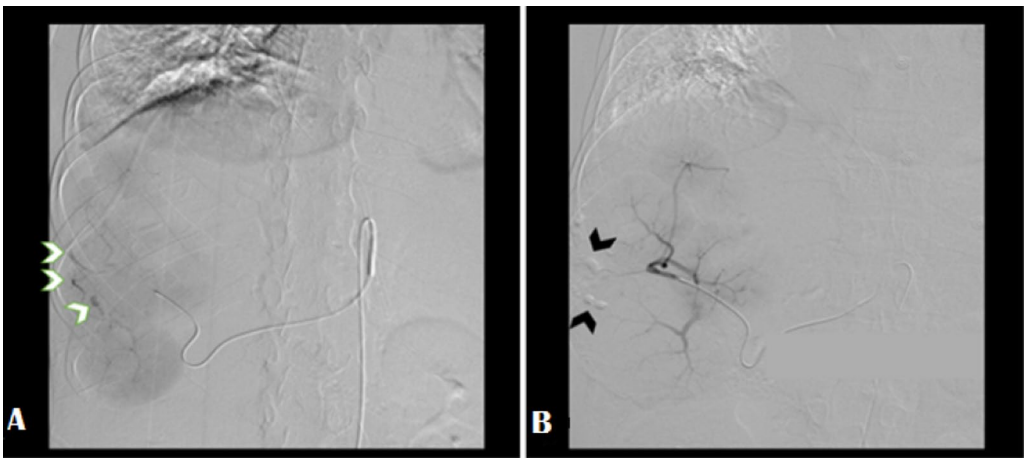

Fig. 3 Digital subtraction angiogram of the splenic artery. **A** Splenic artery run showing small foci of abnormal vascular blush (white arrowheads) in right upper quadrant along splenic artery small branches. These bleeding branches were embolized using Gelfoam embolic. **B** Post-embolization run shows no residual abnormal vascular blush. Faintly visualized deposited Gelfoam can be seen in those areas (black arrowheads)

diagnostic and interventional radiology in diagnosing such patients and identifying the site of bleed especially where endoscopic visualization is not possible and managing the bleeding efficiently.

Abbreviations

EGD: Esophagogastroduodenoscopy; AVSD: Atrioventricular septal defect; VSD: Ventricular septal defect; AV: Atrioventricular; ROSC: Return of spontaneous circulation; ICU: Intensive care unit; NPO: Nil per oral; CECT: Contrast-enhanced computed tomography; MRV: Magnetic resonance venography; SVC: Superior vena cava; IVC: Inferior vena cava; CXR: Chest x-ray.

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Author contributions

MA conceptualized and worked in drafting. NK investigated the data. JS edited the figures. SZS and AR helped in drafting of the manuscript. All authors read and approved the final manuscript. All the authors have agreed to be personally accountable for the author's own contributions.

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Availability of data and materials

Data sharing is not applicable to this article as no datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate

The manuscript has got an ethical review exemption from the Ethical review committee of our hospital as case reports are exempted from review according to our ERC's policy; written informed consent to participate has been obtained from patients Father.

Consent for publication

Written informed consent obtained from patients Father for publication of the case report with the attached images. A copy of the written consent is available for review if needed.

Competing interests

The authors declare that they have no competing interests.

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References

1. Lagrotta G, Moises M (2020) Heterotaxy polysplenia syndrome in adulthood: focused review and a case report. Cureus. https://doi.org/10.7759/cureus.6822
2. Conditions G (2022) Heterotaxy syndrome: MedlinePlus genetics. Medlineplus.gov. https://medlineplus.gov/genetics/condition/heterotaxy-syndrome/. Accessed 31 May 2022
3. Berg C, Geipel A, Kamil D, Knüppel M, Breuer J, Krapp M et al (2005) The syndrome of left isomerism. J Ultrasound Med 24(7):921–931. https://doi.org/10.7863/jum.2005.24.7.921
4. Rahmani M, Salahshour F, Taslimi R, Borhani A, Kianoush S (2016) Extensive gastroesophageal varices in a non-cirrhotic female with left isomerism: a case report and literature review. Iran J Radiol. https://doi.org/10.5812/iranjradiol.34656 (**In Press**)
5. Donnelly A Left sided heterotaxy syndrome (polysplenia). Case study, Radiopaedia.org. https://doi.org/10.53347/rID-89399. Accessed 31 May 2022
6. Shiraishi I, Ichikawa H (2012) Human heterotaxy syndrome. Circ J 76(9):2066–2075. https://doi.org/10.1253/circj.cj-12-0957
7. Kim S (2011) Heterotaxy syndrome. Korean Circ J 41(5):227. https://doi.org/10.4070/kcj.2011.41.5.227
8. Cohen M, Anderson R, Cohen M, Atz A, Fogel M, Gruber P et al (2007) Controversies, genetics, diagnostic assessment, and outcomes relating to the heterotaxy syndrome. Cardiol Young 17(S4):29–43. https://doi.org/10.1017/s104795110700114x
9. Wolla C, Hlavacek A, Schoepf U, Bucher A, Chowdhury S (2013) Cardiovascular manifestations of heterotaxy and related situs abnormalities assessed with CT angiography. J Cardiovasc Comput Tomogr 7(6):408–416. https://doi.org/10.1016/j.jcct.2013.11.008

10. Gayer G, Apter S, Jonas T, Amitai M, Zissin R, Sella T et al (1999) Polysplenia syndrome detected in adulthood: report of eight cases and review of the literature. Abdom Imaging 24(2):178–184. https://doi.org/10.1007/s002619900471

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