Case report

RADIOLOGICAL DIAGNOSIS OF A LEFT ISOMERISM HETEROTAXY ASSOCIATED TO AN ESOPHAGOGASTRIC MOTILITY DISORDER: ABOUT A CASE

ABSTRACT

Situs ambiguous (isomerism) also known as heterotaxy syndrome is an abnormality of lateralization during embryogenesis resulting in atypical anatomy. This is an intermediate configuration between situs inversus and situs solitus with one or more organs in symmetry or even duplicated.

Left isomerism is a splitting of the anatomical configuration of the left side; the right and left sides being identical to the image on the left side, we therefore have polysplenia, an interruption of the inferior vena cava with the azygos/hemiazygos continuation.

We report a particular case of left isomerism of a 12-year-old girl diagnosed by CT scan associating a complete common mesentery with the entire colon located on the left, the small intestine and the stomach distended up the iliac region transposed to the right, a midline liver with transposition of the abdominal vessels and gallbladder, a polysplenia. These anomalies were associated with a megaesophagus probably linked to a motility abdnormality.

Since laterality defects are rare, more data on their anatomical variations could help provide better medical care to this patient population in the future. Their associations with other anomalies, particulary digestives and vasculary ones require particular multidisciplinary attention, the role of the radiologist being central in the management of the multiple variation.

Keywords: Situs ambiguous, heterotaxy, mega esophagus, gastric dilatation

1. INTRODUCTION

Laterality defects are alterations in the left-right axis of the thoraco-abdominal viscera [1]. Their prevalence is estimated at 1.1 per 10,000 live births [2]. A study carried out by Lin et al. in 2014 describing the positioning of the thoraco-abdominal viscera along the left-right axis classifies these abnormalities into 3 main categories, including the situs solitus which is the normal positioning of the viscera; situs inversus totalis (SIT), which involves an inversion of the mirror image of all the viscera; and the ambiguous situs also known as heterotaxy syndrome, which implies that the thoraco-abdominal organs are positioned along the left-right axis in a different way from the situs solitus or the situs inversus totalis [2]. These 3 provisions are considered to be mutually exclusive. In addition, SIT syndrome and heterotaxy syndrome are both laterality defects, but SIT does not usually alter the weakness of the disease for the viscera are always in concordant positions in relation to each other. Therefore, patients with SIT are asymptomatic and the diagnosis is usually incidental [2].

Situs ambigus (isomerism), also known as heterotaxy syndrome, is an abnormality of lateralization during embryogenesis resulting in atypical anatomy. It is an intermediate configuration between the situs inversus and the situs solitus with one or more symmetric, or even duplicated organs. It is defined by

Commented [MA1]: The abstract summarizes the case effectively, the authors can re assign it to have a benefit from some structural refinement. Consider adding specific clinical findings, radiological features, and the implications for clinical practice.

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- You can highlight more detail on the specific clinical challenges or complications associated with left isomerism, especially concerning esophagogastric motility disorders.
- Clarifying the relevance of this association early on, I think would emphasize the novelty of your report.
- Consider restructuring the introduction to improve flow and readability. Starting with the general concept of laterality defects and moving towards more specific aspects (such as heterotaxy syndrome, SIT, and situs ambiguous).
- Please mention the clinical significance of these defects. For example, noting why heterotaxy syndrome is clinically relevant, particularly concerning associated anomalies like megaesophagus!
- Consider expanding on the prevalence or clinical outcomes of heterotaxy syndrome. Including a sentence on the types of complications commonly associated with left isomerism (e.g., cardiac, digestive)!
- A few additional references discussing recent findings or advancements in understanding heterotaxy syndrome would strengthen this section. Since the introduction references Lin et al. (2014), checking for more recent studies that may have expanded on the classifications or prevalence could ensure the report reflects the latest insights.

the symmetry of certain viscera with respect to the sagittal plane: dextro-isomerism or right isomerism when the 2 right and left halves have the morphology of the normal right half (case of asplenia); levoisomerism or left isomerism in the opposite case (polysplenia) [3]. We report a particular case of left isomerism diagnosed by CT scan associating a complete common mesentery with the entire colon located on the left, the small intestine and a distended stomach to the iliac supra transposed to the right, a median liver with transposition of the abdominal vessels and the gallbladder, a polysplenia. However, we noted the association of these abnormalities with the presence of a megaesophagus.

2. PRESENTATION OF THE CASE

This is a patient who was twelve (12) years old at the time of diagnosis, with no particular surgical history, admitted to the pediatric department of Amath Dansokho Regional Hospital in Kédougou, (Senegal) for chronic post-prandial vomiting and weight loss. On clinical examination the general condition was altered by weight loss, the rest was unremarkable. A thoraco-abdomino-pelvic scan (TAP) had been ordered to look for aorto-mesenteric forceps, pyloric stenosis or pancreas divisum. The procedure was performed with a General Electric optima 16-bar CT scan in a fasting patient, supine position and feet first. Three series of acquisitions were performed, one without then with injection of contrast agent at arterial time at the TAP stage, the third at portal time at the abdominal-pelvic (AP) level. The images acquired revealed the following abnormalities: at the thoracic level; there was a diffuse lateral right esophageal dilatation with fluid and air content (Figure 1A and 1B) without suspicious lesions of parietal stenosis, particularly of the cardia; the heart and the large mediastinal vessels were in an anatomical position; there was also a focus of alveolar condensation in the right inferior lobar ventral segment in the pulmonary reconstruction discreetly extended to the middle lobe (Figure 2A), with several parenchymal nodules and micronodules predominantly right in favor of pneumonitis complicating achalasia (inhalation probably). The tracheobronchial tree was normal in appearance. The splits were well placed means two on the right and one on the left (Figure 2B, 2C et 2D). There was no thoracic lymphadenopathy.

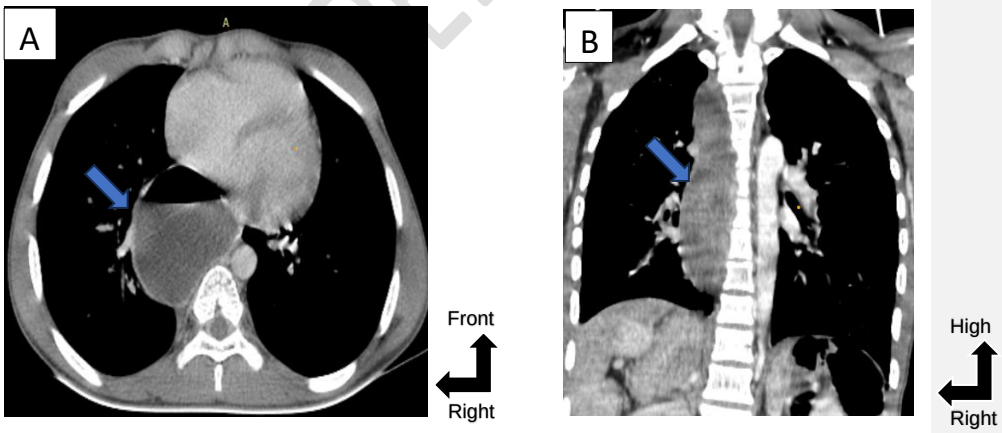

Fig. 1. Chest CT scan with injection in mediastinal reconstruction

A. Axial section: esophageal dilation with hydro-aeric stasis (blue arrow).

B. Coronal section: diffuse esophageal dilatation (blue arrow).

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- For the journal readers, it would be beneficial to include more discussion about differential diagnoses considered initially and how this particular diagnosis was confirmed through imaging and clinical findings.
- Please highlight the potential underlying causes or contributing factors for these symptoms (as post-prandial vomiting and weight loss), including more information on the patient's growth metrics or developmental history might help contextualize the clinical picture, particularly in relation to the motility disorder and its chronic nature.

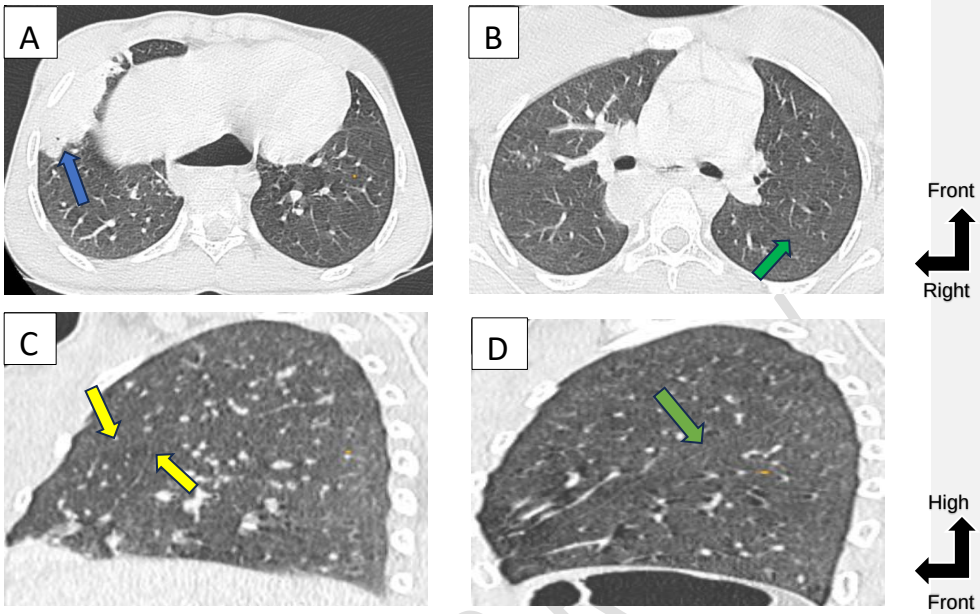

Fig. 2. Chest CT scan in the pulmonary reconstruction

A. In axial section: showing a focus alveolar condensation of the right inferior lobar ventral segment inconspicuously extended to the middle lobe (blue arrow).
B. In axial section: Showing the left fissures (green arrow).
C. In sagittal section: Showing the two right fissures (yellow arrow).
D. In sagittal section: Showing the single left fissure (green arrow).

At the abdominal-pelvic level; The liver has normal size, non-dysmorphic, with regular contours, homogeneous enhancement, in a middle position. The hepatic hilum and gallbladder were transposed to the left, the abdominal aorta was in a normal anatomical position, however there was a transposition of the inferior vena cava to the left of the aorta (Figure 3).

The aorto-mesenteric angle was normal (>20°, no argument in favor of an aorto-mesenteric clamp). The stomach was distended to the iliac supra and transposed to the right (Figure 4) in favor of gastroparesis, there was no suspicious parietal stenosing anomaly, particularly antro-pyloric; the small loops were transposed to the right and in the middle position, the entire colon was transposed to the left consistent with a non-inverted type IA complete common mesentery gastrointestinal tract rotation abnormality (Figure 5A). Multiple splenules of polysplenia were also found in the upper right quadrant (Figure 5B).

The bladder was in semi repletion, presenting spontaneous parietal hyperdensities related to calcifications describing the appearance of a porcelain bladder suggestive of schistosomiasis. The pancreas was seen to be normal. There were no abdominopelvic lymphadenopathy, peritoneal effusions, or bony window abnormalities.

The diagnosis was that of an ambiguous situs (heterotaxy, left isomerism) associated with megaesophagus and gastroparesis gastric distention. An X-ray supplement by oesophageal-gastro-duodenal transit was carried out a few days later, confirming the diffuse esophageal involvement as well as the absence of stenosis by the presence of a satisfactory passage of the barium contrast agent to the small intestine level, suggesting a probable anomaly of oesophageal motility leading to a lack of relaxation, hence the distension (Figure 6). A fibroscopy had been considered after surgical advice.

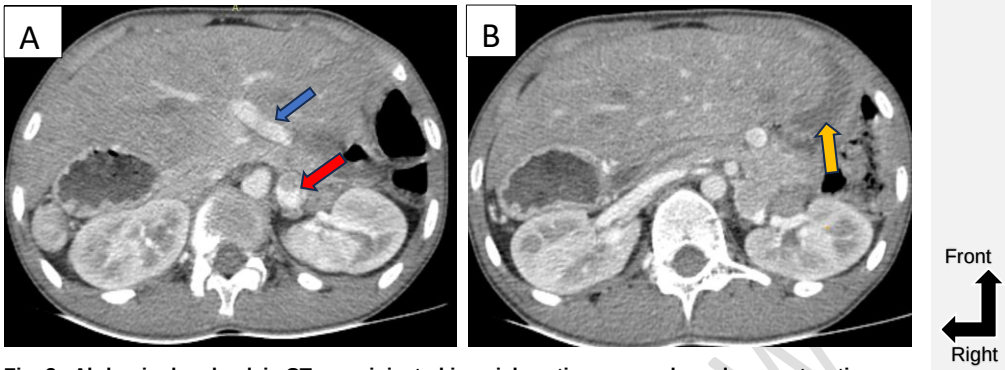

Fig. 3. Abdominal and pelvic CT scan injected in axial section, parenchymal reconstruction

A. Hepatic hilum (trunk with blue arrow), inferior vena cava (red arrow) in a left position compared to the abdominal aorta.

B. Gallbladder (Yellow arrow).

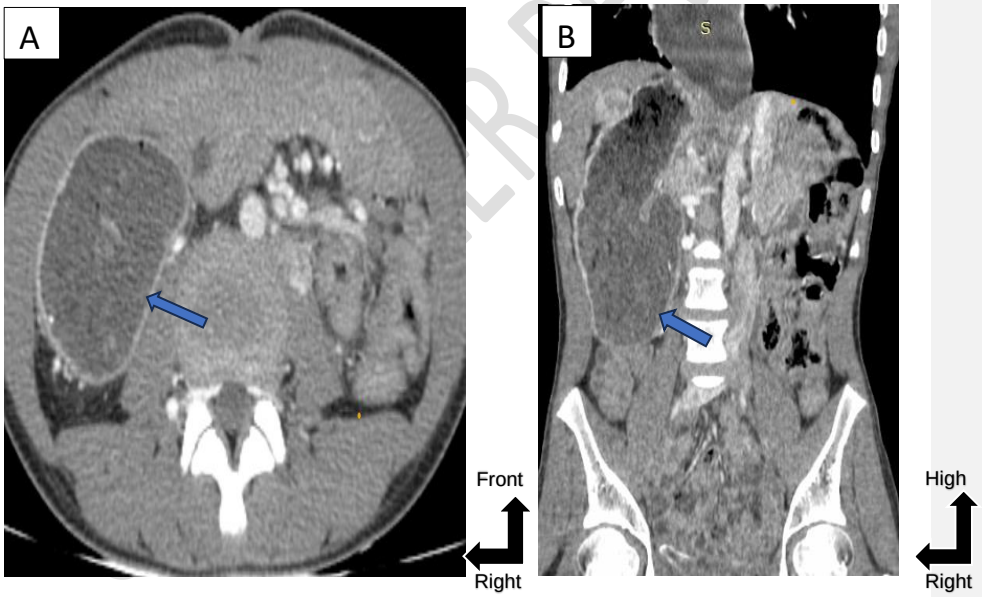

Fig. 4. Injected abdominopelvic CT scan in parenchymal reconstruction

A. Axial section: Dilated stomach transposed to the right with stasis (blue arrow).

B. Coronal section: Gastric dilation with stasis (blue arrow).

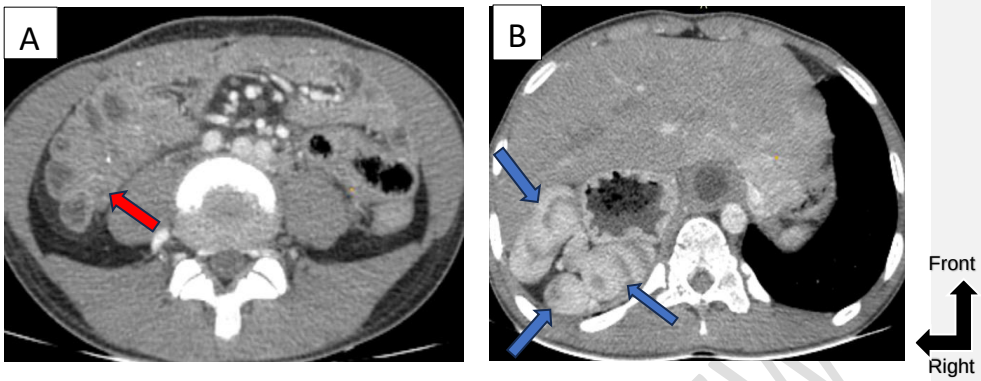

Fig. 5. Abdominal pelvic CT scan injected in axial section and parenchymal reconstruction

A. Gregic portion in the right position (blue arrow) and a colonic portion in the left position (red arrow) in favor of a rotation abnormality of the digestive tract with a complete common mesentery type (type IA not inverted).

B. Multiple splenules (blue arrows) in the upper right quadrant. Note the typical tigerskin enhancement appearance at arterial time.

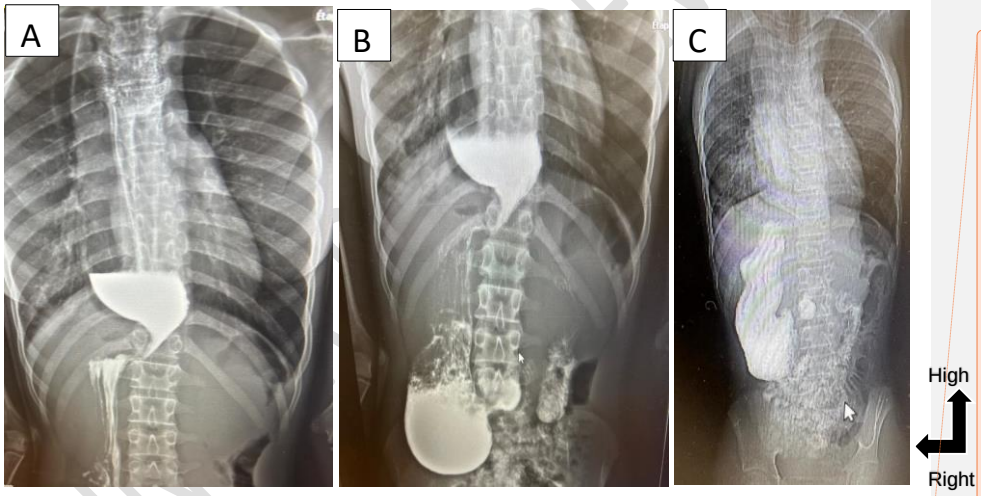

Fig. 6. Gastro-duodenal transit, frontal incidence showing diffuse esophagetric involvement with satisfactory passage of the barium contrast agent to the small level

3. DISCUSSION

Situs ambigus is a rare condition, if we consider all the defects of lateralization [4], women are more affected by it (W>M).

It is defined as a defect in the lateralization of organs, several clinical signs are possible and it can be associated with any type of malformations: cardiac, renal, digestive, etc. but there are two (2) main clinical possibilities:

1- Right isomerism: This would be a doubling of the right side; the thoraco-abdominal anatomical configuration of the right and left sides being identical to the image of the right side in relation to the axis

Commented [MA4]: The authors effectively underscore the uniqueness of this case, emphasizing that this is the first reported instance of left isomerism heterotaxy associated with megaesophagus and gastric distention. This is a valuable contribution to the literature.

- While the discussion provides insights into anatomical anomalies, it would benefit from further elaboration on the clinical implications for patient management. Discussing the potential treatment approaches for megaesophagus in the context of heterotaxy syndrome could add practical value to this report.
- While the discussion references relevant literature, there is an opportunity to integrate recent studies more effectively, especially regarding the clinical management of heterotaxy with gastrointestinal symptoms. This could help position the report within a broader context of current research.
- Consider comparing the findings and management of this case to similar cases, if available, to emphasize its unique contributions.
- Discussing similarities or differences in anatomical findings and clinical outcomes with prior cases would strengthen the discussion.

- The authors can discuss potential areas of further research, particularly given the rarity of this case. For instance, the need for more data on laterality defects with gastrointestinal manifestations could be emphasized.
- The authors should highlight any limitations related to or addressed throughout the workup and case documentation.

of the body, we therefore have a duplication of the organs located on the right in the situs solitus and possible absence of the organs located on the left. Thus, we have: asplenia, a central liver, inferior vena cava on the right and aorta on the left [5].

2- Left isomerism: this would be a doubling of the anatomical configuration of the left side; The right and left sides being identical to the image on the left side, we therefore have a polysplenia, an interruption of the inferior vena cava with the continuation of azygos/hemiazygos [5].

Situs ambigus is in 50 to 100% of cases associated with a congenital heart defect [6]. If the patient does not show signs of life-inconsistent heart disease, he or she may be asymptomatic, having not been diagnosed only incidentally in adulthood [3], often thanks to the medical imaging examinations prescribed for other complaints.

Polysplenia occurs in about 1 in 40,000 live births and usually presents with two (2) or more splenules in the upper left quadrant, instead of a single large spleen [7,8].

Another anatomical abnormality almost as frequently associated with polysplenia is the azygos interruption of the inferior abdominal vena cava in the thorax [9]. Several other anatomical features that are variably associated with polysplenia include dextrocardia, abnormal pulmonary and portal venous return, various congenital heart diseases, bilateral bilobed lungs, midgut malrotation, dorsal pancreatic agenesis, gallbladder agenesis or atresia, and a large median liver [10].

Very few studies have been devoted to ambiguous situs in view of its clinical polymorphism. In Africa, a few rare authors have made a point of talking about the anomalies of the ambiguous situs [11].

We have not been able to find any study talking about the association of ambiguous situs and gastrointestinal abnormality with mega-esophagus type and gastric distension probably linked to a motility disorder. Our case report is the first to describe this unique combination, so it is the first study on the subject. He also emphasizes the importance of radiologists in the management of patients with laterality defects. As these disorders are rare, more data on their anatomical variations could help provide better medical care to this patient population.

The interest of our observation is first of all its association with the presence of a mega-esophagus and a strongly distended stomach to the right iliac supra. Achalasia is a disorder of esophageal motility, characterized by incomplete or absent relaxation of the lower esophageal sphincter and the absence of peristalsis [12]. The phenomenon is rare and affects both men and women of all races.

The average age at diagnosis is over 50 years, but it can be seen in children and young adults [12]. Typical symptoms include progressive dysphagia for solids and liquids (90%), heartburn (75%), regurgitation of undigested food (45%), and respiratory complications, including nocturnal cough and aspiration (20%–40%) [13]. In patients with dysphagia in whom oropharyngeal swallowing is intact, mechanical obstructions should be ruled out by endoscopy or CT scan before motility abnormalities are assessed by esophageal manometry [12]. In the absence of endoscopic lesions, stepped esophageal biopsies should be performed to rule out eosinophilic esophagitis or amyloidosis [14].

The second interest of our observation is the association of left isomerism heterotaxy with a common mesentery. During embryological development, the digestive tract undergoes complex phenomena of reintegration, rotation and adjoining. When these phenomena are incomplete or vicious, they can lead to potentially pathological anatomical situations. In this case, the migration abnormalities of the mesentery: total absence of rotation, complete common mesentery, incomplete common mesentery and reverse rotation in the case of situs inversus. Embryologically, the first rotation takes place before the 10th week of gestation when the primitive intestine is still located outside the abdomen. This rotation places the pre-yolk (hail) portion on the right and the post-yolk portion (colon) on the left; A stop at this stage is the cause of the complete common mesentery. The complete common mesentery is then the result of a 90° halt in intestinal rotation. Thus, the colonic frame on the left and the small intestine on the right; the cecum in the anterior medial position and the superior mesenteric artery to the right of the superior mesenteric vein [15]. The incomplete common mesentery results from a cessation of rotation after two 90° rotations (overall rotation of 180°) --> risk of volvulus of the mesentery (because the root

of the mesentery is short, the right iliac fossa is uninhabited with cecum in the upper middle or subhepatic position.

Preferred imaging modalities for laterality defects

There is no optimal imaging modality for laterality defects, as each type has its own advantages and disadvantages for different body systems [16]. Since heterotaxed patients often have symptoms of congenital heart disease, the first imaging they may receive is a chest X-ray. Similarly, patients with abdominal pain may receive an abdominal x-ray (abdominal plain film) during their initial workup. X-rays can identify major anatomical defects such as a median heart and liver, symmetrical bronchial branching, or an upright stomach, but it does not provide a more detailed description of the anatomy of the heart chambers, pancreas and spleen, or intestines [16].

Other imaging modalities that should be included in the initial workup of patients include echocardiography, abdominal ultrasounds, and upper gastrointestinal series [17]. Echocardiography can diagnose and characterize congenital heart disease, while abdominal ultrasounds assess intra-abdominal contents. Similarly, upper gastrointestinal series may exclude malrotation of the intestines, which predisposes patients to volvulus. Patients also commonly receive contrast CT imaging, which provides information about vascular anatomy [9,10].

4. CONCLUSION

The ambiguous situs is a real anatomical curiosity because of its numerous clinical and especially radiological presentations. Our case report is the first to describe the unique combination of heterotopia of left isomerism, common mesentery, megaesophagus and gastric distension on oesogastric motility abnormality. Since laterality defects are rare, more data on their anatomical variations could help provide better medical care to this patient population in the future. Their associations with other abnormalities, particularly digestive and vascular, require special multidisciplinary attention, the role of the radiologist being central in the management of these multiple anatomical variations.

CONSENT

As per international standard or university standard, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

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Commented [MA5]: No need to highlight such subheadings in the discussion section!

Commented [MA6]: For the readers, the authors can highlight :

- The radiological evaluation in recognizing rare anatomical variations associated with laterality defects.
- How this case can affect future clinical practice, especially in the early recognition and management of similar cases, would provide a stronger closure to the discussion.
- Consider summarizing the primary learning points from this case to reinforce the report's educational impact for clinicians who may encounter similar presentations.

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- An acknowledgment section should be submitted.
- The authors should highlight a conflict of interest.

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- The references are generally appropriate but could be updated to include more recent studies, especially if new developments on heterotaxy syndromes or esophageal motility disorders are available.
- Cross-check the references for accuracy, particularly regarding publication dates and titles, to ensure precision.

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