Case report

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

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 case of left isomerism of a twelve -year-old girl diagnosed by CT scan associated with entire colon and complete common mesentery located on the left, small intestine and stomach distended up the iliac region transposed to the right, a midline liver with transposition of the abdominal vessels and gallbladder, and a polysplenia. These anomalies were associated with a megaesophagus probably linked to a 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 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], with an estimated prevalence of 1.1 per 10,000 live births [2]. In their study describing the positioning of the thoraco-abdominal viscera along the left-right axis, Lin et al. classify these abnormalities into three main categories, including the situs solitus which is the normal positioning of the viscera; the situs inversus totalis (SIT), an inversion in which all viscera mirror the normal layout; and the ambiguous situs, also known as heterotaxy syndrome, in which the thoraco-abdominal organs are positioned differently from either situs solitus or situs inversus totalis [2]. These three provisions are mutually exclusive. Additionally, while SIT syndrome and heterotaxy syndrome are both considered as laterality defects, SIT typically does not compromise organ function, as the viscera remain in concordant positions relative to one another. Consequently, patients with SIT are asymptomatic, with diagnosis usually being incidental [2].

Situs ambigus (isomerism), also known as heterotaxy syndrome, is a lateralization abnormality during embryogenesis that results in atypical anatomy. It represents an intermediate configuration between the situs inversus and the situs solitus with one or more symmetrical, or even duplicated organs. Situs

ambigus is defined by the symmetry of certain viscera relative to the sagittal plane : dextro-isomerism or right isomerism occurs when both the right and left halves display the morphology of the normal right half (as in case of asplenia) , while levoisomerism or left isomerism occurs in the opposite case (polysplenia) [3]. We report a case of left isomerism diagnosed by CT scan, revealing a complete common mesentery, with the entire colon located on the left, the small intestine and a distended stomach distended up the iliac region transposed to the right, a median liver with transposition of the abdominal vessels and gallbladder, and a polysplenia. Additionally, these abnormalities were assioted with the presence of a megaesophagus which the main complication can be a bronchopneumonia due to false swallowing, which is manifested by cough, dyspnea, purulent discharge, sometimes hyperthermia.

2. PRESENTATION OF THE CASE

We present the case of a twelve-year-orld patient 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 remainder of the examination was unremarkable. There was not enough information on the patient's growth metrics or developmental history. No dietary particularities were noted in particular no allergies.

A thoraco-abdomino-pelvic scan (TAP) was ordered to investigate some clinical differential diagnoses like superior mesenteric artery syndrome , pyloric stenosis or pancreas divisum. The procedure was performed using a General Electric optima 16-row CT scan in a fasting patient, supine position and feet first. Three series of acquisitions were performed: non-contrast and post-contrast in arterial phase scanning the region of thorax, abdomen and pelvis, and post-contrast in portovenous phase scanning abdomen and pelvis regions. The acquired images revealed the following abnormalities: at the thoracic level; there was a diffuse lateral right esophageal dilatation containing fluid and air content (Figure 1A and 1B) without any suspicious lesions of parietal stenosis, particularly at the cardia; the heart and major mediastinal vessels were anatomically positioned. In addition, there was a focus of alveolar consolidation 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 suggestive of pneumonitis complicating achalasia (likely due to aspiration). The tracheobronchial tree appeared normal. The fissures were well-positioned, with two on the right and one on the left (Figure 2B, 2C et 2D). No thoracic lymphadenopathy was observed.

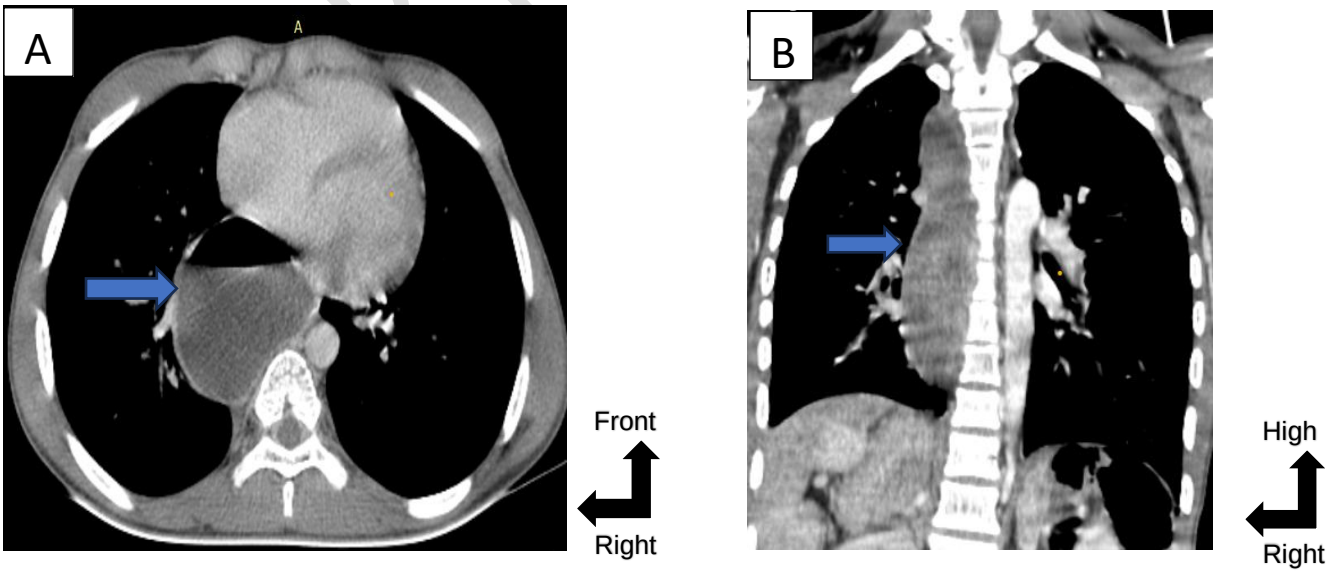

Fig. 1. Chest CT scan with injection in mediastinal reconstruction

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

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

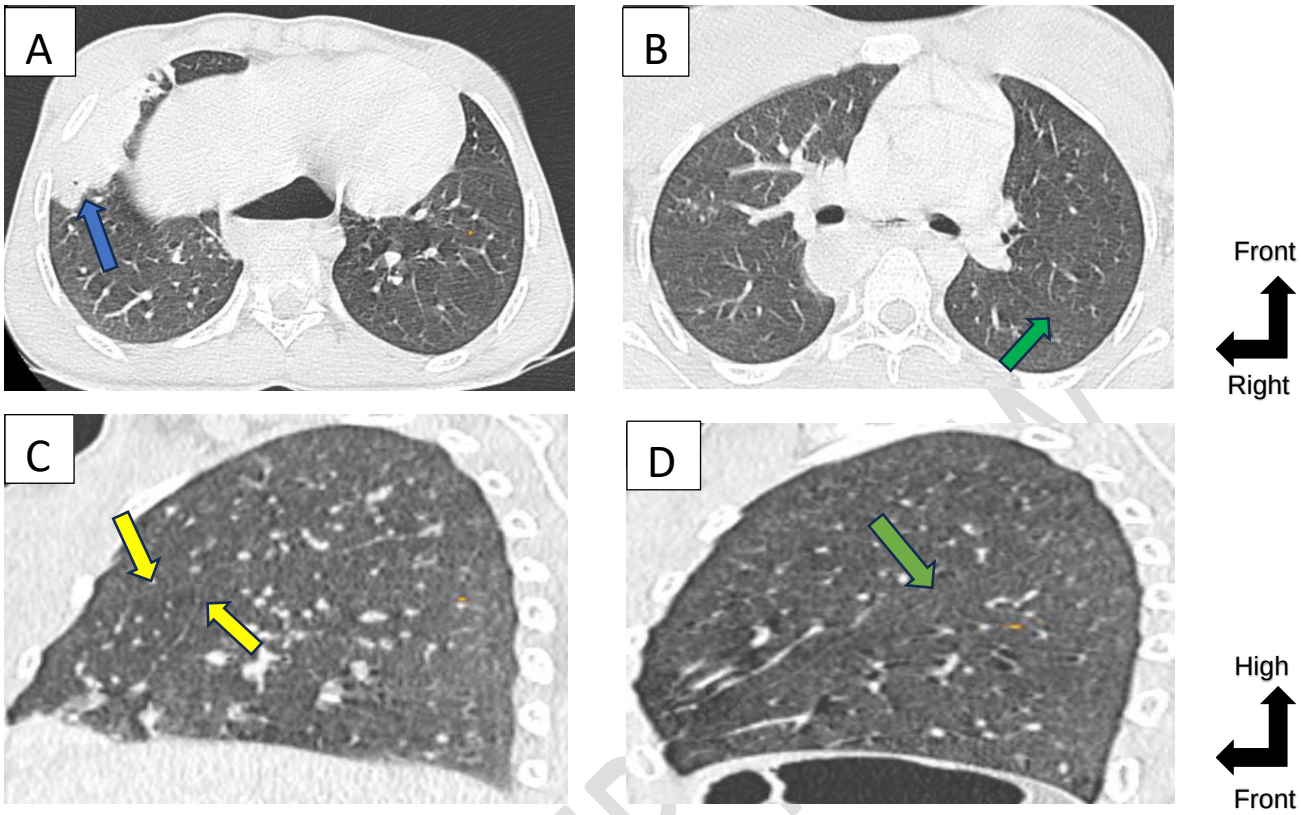

Fig. 2. Chest CT scan in the pulmonary reconstruction

A. In axial slice: showing a focus alveolar consolidation of the right inferior lobar ventral segment inconspicuously extended to the middle lobe (blue arrow).
B. In axial slice: Showing the left fissures (green arrow).
C. In sagittal slice: Showing the two right fissures (yellow arrow).
D. In sagittal slice: 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°, with no evidence of an aorto-mesenteric clamp). The stomach was distended up the right iliac region and transposed to the right (Figure 4) suggestive of gastroparesis. There was no suspicious parietal stenosing anomaly, particularly at the antro-pyloric; the small loops were transposed to the right and in the middle position, and 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 ambiguous situs (heterotaxy, left isomerism) associated with megaesophagus and gastroparesis with gastric distention.

A supplementary X-ray with oesophageal-gastro-duodenal transit was performed a few days later, confirming the diffuse esophageal involvement and the absence of stenosis , as indicated by the satisfactory passage of the barium contrast agent to the small intestine, 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.

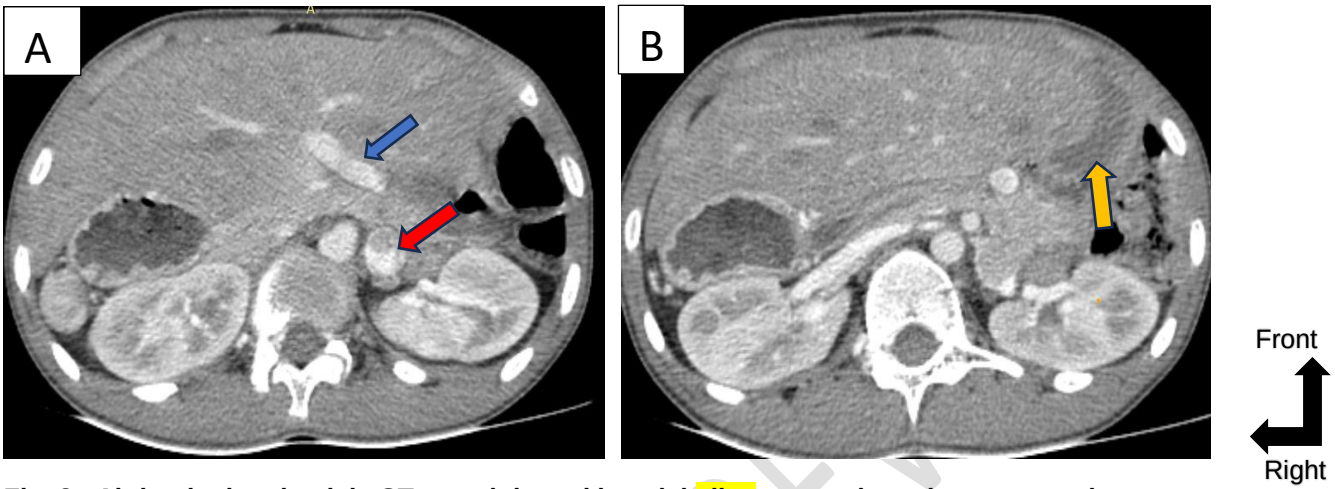

Fig. 3. Abdominal and pelvic CT scan injected in axial slice, parenchymal reconstruction

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).

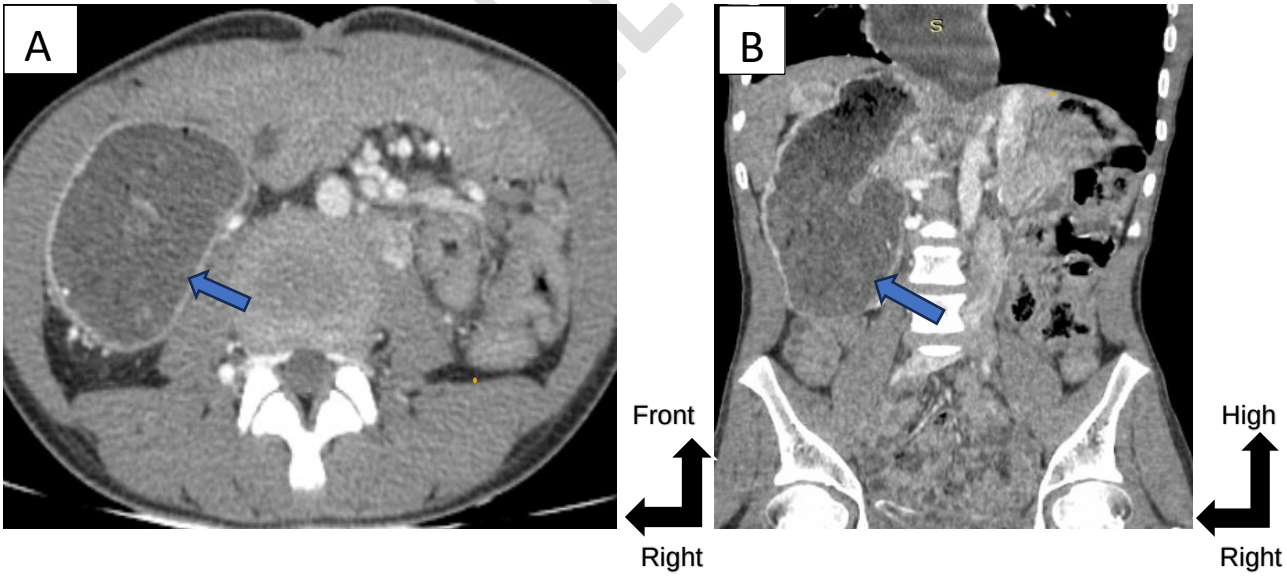

Fig. 4. Injected abdominopelvic CT scan in parenchymal reconstruction

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

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

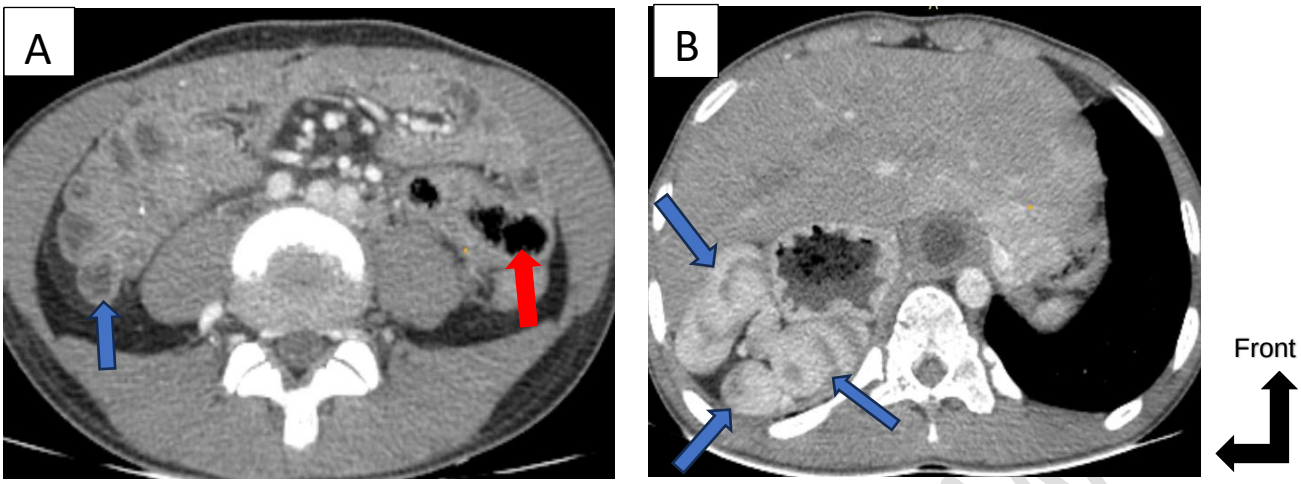

Fig. 5. Abdominal pelvic CT scan injected in axial slice and parenchymal reconstruction

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.

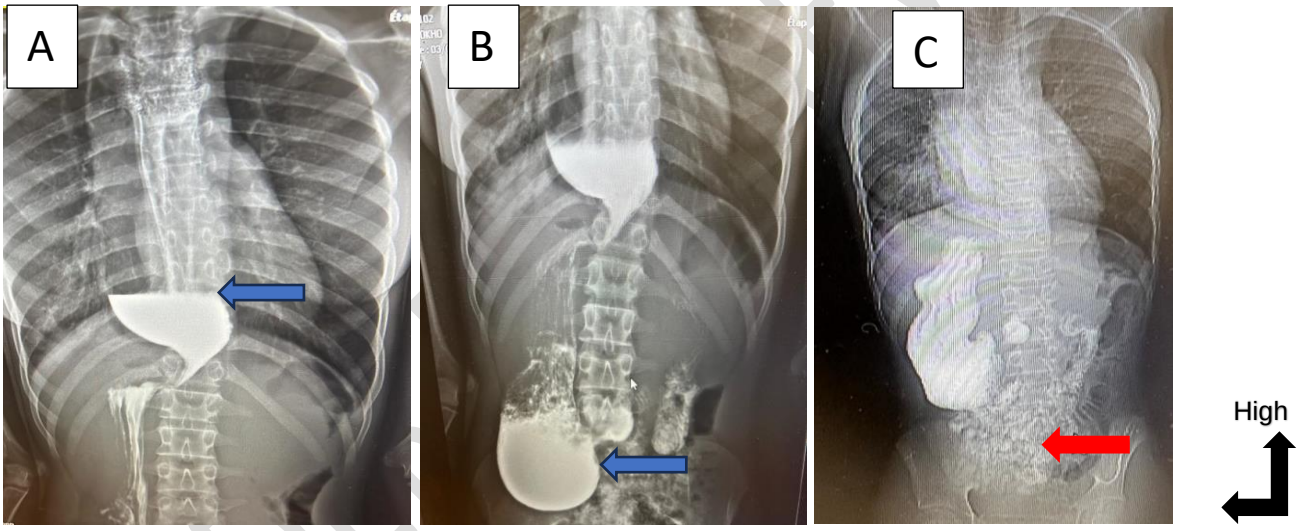

Fig. 6. Gastro-duodenal transit, frontal incidence showing diffuse esophagetric and gastric involvement (blue arrows) with satisfactory passage of the barium contrast agent to the small level (red arrow)

3. DISCUSSION

Situs ambigus is a rare condition, when considering all the lateralization defects [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 refers to a duplication 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 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. There is therefore a need for more data on laterality defects accompanied by gastrointestinal manifestations. Data ranging from clinical manifestations (including complications) to radiological signs.

The interest of our observation is first of all its association with the presence of a mega-esophagus and a strongly distended stomach up the right iliac region. 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.

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

Radiology plays a central role in the diagnosis of rare anatomical variations associated with laterality defects. 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 of a 12-year-old girl presenting with clinical signs associating chronic postprandial vomiting and weight loss. 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.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Authors hereby declare that NO generative AI technologies such as large language models (ChatGPT, COPILOT, etc) and text-to-image generators have been used during writing or editing of this manuscript.

CONSENT

As per international standard or university standard, patient and parents 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).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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REFERENCES

1. Antony, D., Gulec Yilmaz, E., Gezdirici, A., Slagter, L., Bakey, Z., Bornaun, H., et al. Spectrum of genetic variants in a cohort of 37 cases of laterality defects. Front genet. 2022;13 doi: 10.3389/fgene.2022.861236. French.

2. Lin, AE., Krikov, S., Riehle-Colarusso, T., Frías, JL., Belmont, J., Anderka, M., et al. Laterality anomalies in the National Congenital Anomalies Prevention Study (1998–2007): birth prevalence and descriptive epidemiology. Am J Med Genet A. 2014;164(10):2581–2591. doi: 10.1002/ajmg.a.36695. French.

3. Andres, A., Maria, N.M., Margarita, F. Heterotaxy Syndrome in a Young Adult. American Journal of Clinical Medicine Winter.2012; 9(1): 36-44.

4. Sutherland, M.J., Ware, S.M. Left-right asymmetry disorders: heterotaxy and situs inversus. Am J Med Genet C Semin Med Genet .2009 Nov 15;151C (4): 307-17. French.

5. Winer-Muram, H.T., Tonkin, I.L. The spectrum of heterotaxic syndromes. RadiolClin NorthAm. 1989;27(6): 1147-70.

6. El guindi, W., Dreyfus, M., Carles, G., Lambert, V., Herlicoviez, M., Benoit, G. The contribution of 3D ultrasound in heterotaxy syndromes: about four cases and review of the literature. Journal of Gynecology Obstetrics and Reproductive Biology. 2012;41(5):489-496. French.

7. Shiraishi, I., Ichikawa, H. Human heterotaxy syndrome. Circ J. 2012;76(9):2066–2075. doi:10.1253/circj.cj-12-0957. French.

8. Agarwal, R., Varghese, R., Jesudian, V., Moses, J. Heterotaxy syndrome: associated congenital heart defects and management. Indian J Thorac Cardiovasc Surg. 2020; 37 (S1): 67–81. doi: 10.1007/s12055-020-00935-y. French.

9. Malki, M.C., Outznit, M., Mechhor, S., Bouibaouen, B., Nkurunziza, L., Elbacha, H., et al. Adult-onset polysplenia syndrome: a case report. PAMJ One Health. 2022; 41 doi: 10.11604/pamj.2022.41.67.29014. French.

10. Gayer, G., Apter, S., Jonas, T., Amitai, M., Zissin, R., Sella, T., et al. Adult-Detected Polysplenia Syndrome: Report of Eight Cases and Review of the Literature. Abdom Imaging. 1999; 24 (2): 178–184. doi: 10.1007/s002619900471. French.

11. Basse, I., Mbengue, M., Coly, N.F., Sibabi, A.B., Diaby, F., N´Diaye, N. et al. Childhood Heterotaxy, from Diagnosis to Management: A Report of 3 Cases. Black African Medicine. 2021: 531–534. French.

12. Boeckxstaens, G.E., Zaninotto, G., Richter, J.E. Achalasia. Lancet 2014;383:83-93 13. Pandolfino, J.E., Gawron, A.J. Achalasia: a systematic review. JAMA 2015;313:1841-52 14. P. Castro Soares, M., Drepper, R., Grignoli, P., Bichard, J.L, Frossard. Achalasia: which therapy to choose in 2015 Rev Med Suisse 2015; 11:1587-91. French.

15. Traoré, A.A., Mvumbi, K.F., Ly, S., Alaoui, L.Y., Boubbou, M., Maaroufi, M. et al. Computed tomographic finding of a complete common mesentery due to perforation of acute appendicitis. Pan Afri Med J. 2017;27:3. French.

16. Loomba, R., Shah, P.H., Anderson, R.H., Arora, Y. Radiological considerations in heterotaxy: the need for detailed anatomic evaluation. Cureus. 2016;8(1):e470.doi:10.7759/cureus.470. French.

17. Applegate, K.E., Goske, M.J., Pierce, G., Murphy, D. Situs revisited: imaging of the heterotaxy syndrome. Radiographics. 1999;19(4):837-852.doi:10.1148/radiographics.19.4. g99jl31837. French.