



Thoracoabdominal duplication accompanied by intestinal malrotation: a case report and literature review of a rare congenital anomaly in an infant

Qusai Mashlah, MD^{a,b}, Omar Al Laham, MBBS^{a,*}, Hajar Odah Bashi, MD^{a,b}, Rahaf Sharaf Aldeen, MD^{a,c,d}, Sami Alashi, MD^a, Mohammmd Abdulkader, MD^{a,b}

Introduction and importance: Thoracoabdominal duplication and intestinal malrotation are extremely rare congenital alimentary tract anomalies that can manifest in any segment of the gastrointestinal tract. Still, tubular duplications are an even rarer subset of alimentary tract duplications. Misdiagnosis could occur and this will yield devastating ramifications. Therefore, consideration in the clinical settings is warranted to aid in conducting timely therapeutic interventions.

Case presentation: In this article, we illustrate the overwhelmingly rare occurrence of thoracoabdominal duplication coexistent with intestinal malrotation in a 7-month-old male whose primary complaint was chronic dyspnoea since birth that progressed to involve cough and fever. Imaging analysis revealed a significant intrathoracic fluid-filled cyst.

Clinical discussion: The intestinal malrotation was treated through Ladd's procedure, and surgical excision of the duplicated segments was accomplished. The subsequent analysis of the resected specimens via means of histopathology utilizing Hematoxylin and Eosin dyes established the definitive diagnosis of a foregut duplication cyst.

Conclusion: Thoracoabdominal duplication is one of the most crucial topics in the field of Paediatric Surgery. It is exceptionally rare in occurrence, and the scarcity of available resources that document and describe this topic is evident in the published literature. The authors must opt to document, study, and broaden awareness regarding this life-threatening pathology so that they can circumvent the resultant complications by means of early detection and the performance of apt surgical interventions. Upon careful review of the available literature, we can state that ours is the first-ever case documented from their country regarding this topic and this co-incidence.

Keywords: abdominal surgery, alimentary tract duplications, case report, congenital anomaly, foregut duplication cyst, pediatric surgery

Introduction

Gastrointestinal tract duplication is an exceedingly rare congenital pathology that is estimated to be affect one out of every 4500 live births^[1]. They could extend via the diaphragm as this was first documented by Valle and White in 1946 and this anomalous occurrence comprises merely 2% of all cases of alimentary tract duplications^[2].

^aFaculty of Medicine, ^bDepartment of Pediatric Surgery, Children's University Hospital, ^cDepartment of Surgery, Al-Mouwasat University Hospital and ^dDepartment of Surgery, Al Assad University Hospital, Damascus University, Damascus, (The) Syrian Arab Republic

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*Corresponding author. Address: Faculty of Medicine, Damascus University, Damascus, (The) Syrian Arab Republic. E-mail: 3omar92@gmail.com (O. Al Laham).

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HIGHLIGHTS

- Gastrointestinal tract duplication is an exceedingly rare congenital pathology that is estimated to be affect one out of every 4500 live births.
- Extending via the diaphragm, as was first documented by Valle and White in 1946, comprises merely 2% of all cases of alimentary tract duplications.
- Intestinal malrotation is the failure of the foetal intestines to achieve a complete 270° counterclockwise rotation cycle around the superior mesenteric axis, and its incidence rate is 0.2% of all live births.
- Treatment for both conditions is optimally achieved via surgery in the form of Ladd's procedure for malrotation and resection of the duplication cysts for alimentary tract duplication.
- Ours is the first-ever documented case from our country and the 76th one worldwide of Thoracoabdominal duplication.

Such alimentary tract duplications are extremely uncommon congenital abnormalities that can originate anywhere along the gastrointestinal tract. The majority of these occurrences are successfully diagnosed during the first two years of life. Moreover, cystic duplications are more prevalent, whereas tubular duplications are largely less common^[3].

In terms of the time of clinical presentation, this anomalous development typically exhibits symptoms during the first few months of the first year of life. However, accurately diagnosing it in the clinical setting poses a challenge since the corresponding symptoms are non-specific^[4].

With regard to our other topic; intestinal malrotation, the clinical definition of it is the failure of the intestines of a foetus to achieve a complete 270° counterclockwise rotation cycle around the superior mesenteric axis by the time of 10 gestational weeks^[5]. The recorded incidence rate is estimated to be around 0.2% of all live births. Furthermore, this pathology is consequently immensely rare and the result of this anomalous embryogenic development yields both functional as well as anatomical anomalies of the small and large intestines^[6].

It is vital to emphasize the need for establishing an early diagnosis because it is the keystone to reducing the high morbidity and mortality that are resultant from the subsequent complications, such as intussusception, volvulus, herniation^[7] and even death.

Following comprehensive review of the published literature, we can deduce that ours is the first-ever case documented from our country and the 76th case worldwide regarding this topic and this co-occurrence. Moreover, it is also the first-ever documented one of a thoracoabdominal duplication concomitant with intestinal malrotation.

The work has been reported in line with the SCARE criteria and the revised 2020 SCARE guidelines^[8].

Presentation of case

Patient information

We report a rare case of a 7-month-old Middle Eastern male infant who presented with a history of dyspnoea since birth. At the age of 4 days, an echocardiogram revealed the presence of an intrathoracic fluid-filled cyst, which was not adequately addressed by the parents. Since the beginning of the fourth month, the child appeared to be underweight. Additionally, there was a delayed psychomotor development. The infant subsequently developed fever, cough and progressive dyspnoea without cyanosis a week before admission. The gestational age is unknown, and there were no notable findings during prenatal monitoring.

The remaining systemic review was unremarkable.

His previous medical, surgical, allergic, and drug histories are negative.

Clinical findings

We started our physical examination of the patient by recording the vital signs and they were normal except for mild tachypnea.

Upon inspection, the patient did not exhibit any cyanosis, pallor, shallow breathing, or sternal retraction. Inspection of the abdomen showed a symmetrical movement with respiration, the umbilicus was central without deviation, and no distention or skin discolorations were seen (i.e. Spider naevi, caput medusa, icterus, or hypo/hyperpigmentation).

No guarding or tenderness were elicited upon abdominal palpation or percussion.

Lastly, the auscultatory findings of the chest revealed decreased breath sounds over the right side but they were normal over the

abdomen. The remainder of the physical evaluation did not yield any other findings.

Diagnostic assessment

The preoperative radiological evaluation was initiated by performing a simple chest X-ray image which revealed a mass effect on the right side, atelectasis of the right lung and a left mediastinum shift (Fig. 1). This was followed by a pleural ultrasonography scan which indicated a small amount of effusion on the right side, and a cystic thoracic mass encompassing hyperechoic fluid contents, which is non-compressible but pushes the heart to the left. The heart was otherwise normal.

Further complex imaging modalities were necessary to obtain a more detailed view of the situation. Therefore, a Computed Tomography (CT) scan was done and it demonstrated a large (9 cm) cystic lesion in the posterior mediastinum extending to the right chest. The lesion's wall is relatively thick and contains a gas-fluid level with a left shift of the mediastinum and right pleural effusion (Fig. 2A–F).

A complete laboratory workup was also done. The laboratory panel revealed a white blood cell count of (10 900 cells/mm³). The haemoglobin level was (7.6 g/dl), and the platelets (855 000 cells/mm³). The C-reactive protein level was slightly elevated at (2.4 mg/dl). All other laboratory values were within normal ranges.



Figure 1. Preoperative chest X-Ray revealing a mass effect on the right side, right lung atelectasis, and a left shift of the mediastinum.

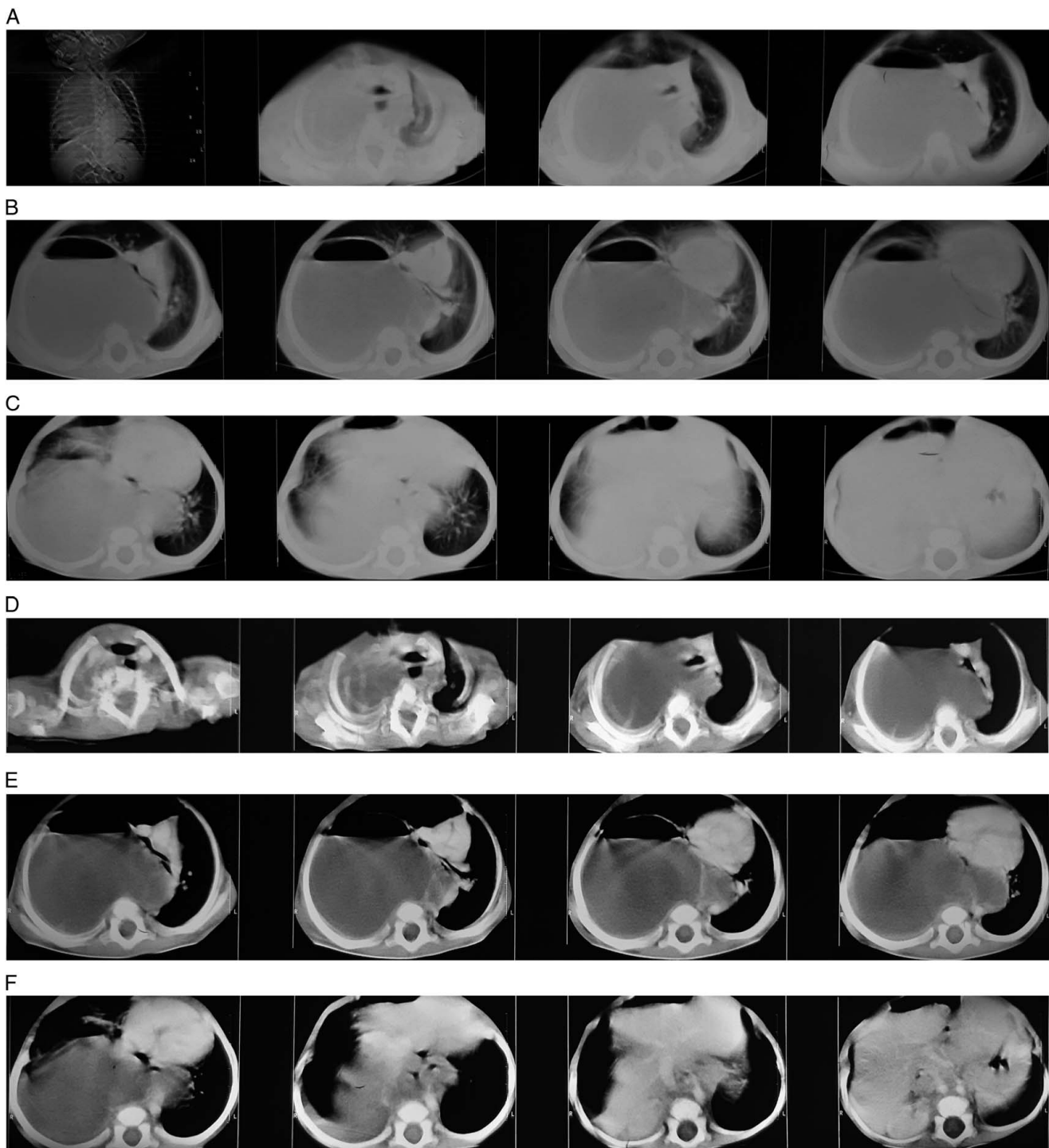


Figure 2. (A–F): Preoperative Computed Tomography scan of the chest demonstrating a large (9 cm) cystic lesion in the posterior mediastinum extending to the right chest. The lesion's wall is relatively thick and contains a gas-fluid level with a left shift of the mediastinum and right pleural effusion.

Our team reached the decision to perform a curative surgical intervention after multidisciplinary consultations. To prepare him for surgery, he received a blood transfusion and had adequate intravenous cannulas set up to administer the suitable fluid infusions and antibiotics.

Therapeutic intervention

An elective surgical intervention was deemed appropriate to treat this clinical picture. The surgery was carried out at our tertiary university hospital under the umbrella of general anaesthesia. The procedure was accomplished by a Pediatric Surgery specialist and

a senior resident as a first assistant with 16 and 6 years of surgical experience, respectively.

A right lateral thoracic incision was made, revealing a large cystic structure that filled the chest cavity. The cyst was dissected from the original oesophagus towards the neck where the upper end was firmly attached to the body of the vertebra. Further investigation revealed that it was a thoracoabdominal duplication extending into the abdomen through its own hiatus without direct contact with the intestinal tract. A decision was made to perform a laparotomy via a midline incision. During the procedure, it was found that the cyst passed behind the liver and connected with the third portion of the duodenum without sharing the same mesentery. The cyst was then completely excised. Additionally, we saw the presence of a concomitant intestinal malrotation and the presence of Ladd's bands and marked adhesions. As a result, we dissected Ladd's bands, resected the appendix, folded the small bowel into the right side of the abdominal cavity, placed the colon into the left side, and carried out meticulous adhesiolysis. This is otherwise known as Ladd's Procedure.

Further in-depth exploration of the chest and abdomen was done and no other findings were marked. We would like to note, that we did not see, intraoperatively, any evidence of pulmonary hypoplasia.

The resected specimens were immediately sent to a specialized histopathology laboratory for further analysis and to set a definitive diagnosis.

This analysis via utilization of Hematoxylin and Eosin (H&E) staining reported the presence of mixed epithelial lining, squamous, gastric mucosa, ciliated pseudostratified columnar and columnar mucosa overlying a double layer of smooth muscle. It also contains solid foci of pancreatic tissue (Fig. 3A, B).

Postoperatively, a chest X-ray was performed with the purpose of confirming that there was no evidence of severe pulmonary hypoplasia on the right side. However, it revealed signs of lobar consolidations (Fig. 4) which was treated medicinally in the intensive care unit.

The patient underwent extubation on the second postoperative day and commenced oral feeding on the fourth postoperative day with good tolerance. However, on the fifth postoperative day, the patient presented with severe respiratory distress, requiring emergent nasotracheal intubation. He was provided with the necessary urgent medical care. However, the patient's clinical status rapidly and progressively deteriorated, ultimately resulting in death due to respiratory failure on the 11th postoperative day.

Discussion

In 1950, Ladd & Gross were the pioneers who described the duplications of the gastrointestinal tract as follows: "Spherical or tubular formations that can occur from the mouth to the anus, having contact with the normal digestive tract, communicative or not, with a smooth musculoskeletal and a digestive-type mucosa"^[9]. In turn, duplications that are termed "Thoracoabdominal duplications" are an extremely rare subtype and comprise merely 2% of all types of alimentary tract duplications. A percentage of which reaching almost 60% of occurrences could possess a direct connection with the digestive system^[10].

With regard to gender-specific dominance, it has been found that there is a slight male-to-female predominance ratio of 1.2:1^[11-13].

In terms of site of occurrence of this anomaly, it could principally be the oesophagus^[14], the stomach alone or in addition to

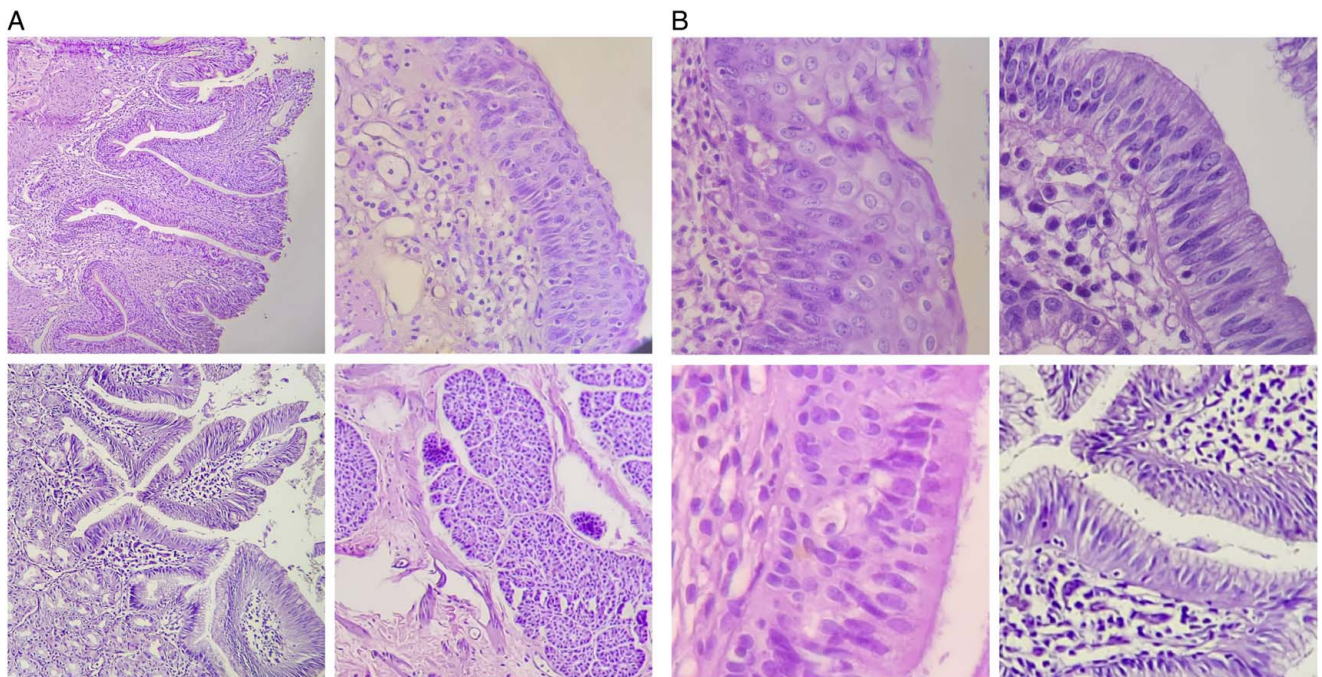


Figure 3. (A and B) Histopathological analysis of the resected cystic lesion via Hematoxylin and Eosin staining which revealed the presence of mixed epithelial lining, Squamous, gastric mucosa, ciliated pseudostratified columnar and columnar mucosa overlying a double layer of smooth muscle. It also contains solid foci of pancreatic tissue.



Figure 4. Chest X-ray on the third postoperative day aimed to clarify that there were no signs of severe pulmonary hypoplasia on the right side. Nevertheless, we can denote the presence of pulmonary lobar consolidation that was medically managed.

the oesophagus^[12], and moreover, the duodenum^[13]. When originating from an anatomical location within the thorax, this portion of the duplication archetypally extends to the level of a malformation in the cervical/thoracic spine. Nonetheless, when said segment crosses the diaphragm in much rarer cases, it is usually seen to occur via a deep orifice in two thirds of cases. In the minority of cases, this crossing occurs via the aortic or oesophageal orifices^[14] as was the situation in our case.

As for the age of clinical presentation, such an anomaly is in almost 75% of cases detected within the first year of life due to the manifestation of different respiratory system signs as a result of physical compression effect of the lesion to the nearby respiratory tract. Still, they could also be diagnosed as a result of digestive system symptoms exhibited as a result of the production of acid contents from atopic gastric mucosa^[11,12]. It is worth noting that this pathology is seldom seen in older individuals^[13].

In circumstances where symptoms are elicited, they include fever, stunting as a result of weight stagnation, and respiratory system signs and symptoms due to the aforementioned physical compression effects^[13]. On the other hand, probable abdominal manifestations of this ailment habitually include gastrointestinal bleeding, alternating pain, and peritonitis as a result of the life-threatening perforations^[12].

Establishing a preoperative diagnosis utilizing radiological assessment tools poses a considerable challenge as it is highly challenging to accomplish^[14]. Moreover, regular thoracic and abdominal X-rays are predictably advantageous in indicating homogenous fluid opacities in the posterior mediastinum. This is frequently found on the right side, posterior to the heart, and inferiorly located lesions accompanied with visualized defects in thoracic and cervical vertebrae^[15].

In terms of ultrasonography scanning, abdominal and chest ultrasound examinations classically expose findings consistent with the cystic nature of the lesion and demarcate its location and extent as much as possible^[16].

With regard to the therapeutic treatment of thoracoabdominal duplications, it is agreed upon that they ought to be resected due to their dangerous potential complications if left existing^[17,18]. To this date, the cornerstone for treatment of thoracoabdominal duplications remains surgery whereby the resected specimens are then sent for the proper histopathological analysis that establishes a definitive diagnosis^[4].

In terms of complications that we attempt to circumvent, the possible existence of heterotopic acid-secreting gastric mucosa and possible peritonitis due to perforation are the main complications that are avoided by performing utter surgical removal of the lesions^[17]. Upon review of the available literature, all documented cases of thoracoabdominal duplications since their earliest depiction by Valle & White have been managed either with thoracotomy in concomitance with laparotomy or with a thoracophreno-laparotomy^[19]. Generally, it takes two discontinuous incisions and may be performed in one or two stages. Surgical resection both anatomical segments of the duplications; Abdominal and thoracic during in the same procedure has been proposed as an alternative approach to attempt in averting any potential complications^[20].

Table 1

The following Table provides a summary of the published cases in the literature of thoracoabdominal duplication.

Study author	No. cases	Year	Reference number
Gross RE	3	1952	[21]
Shepherd M	1	1965	[22]
Lawson R	1	1968	[23]
Fitzgibbons R	1	1980	[24]
Schwöbel M	1	1982	[25]
Aubrespy P	2	1984	[17]
Pokorny W	25	1984	[11]
Giacoia G	1	1984	[26]
Holcomb G	3	1989	[3]
Wolf Y	1	1990	[27]
Pintér A	2	1992	[28]
Thornton J	1	1992	[29]
Ayadi K	1	1994	[30]
Stringer M	6	1995	[31]
Savci G	1	1997	[32]
Karmak I	2	2000	[33]
Danzer E	1	2001	[34]
Bhat N	1	2001	[10]
Bhat N	6	2001	[35]
Nelms C	1	2002	[36]
Martinez-Ferro M	1	2005	[19]
Hishiki T	1	2006	[37]
Zhang J	1	2010	[38]
Bui T	1	2013	[18]
Moralioğlu S	1	2014	[20]
Jehangir S	3	2015	[39]
Ravina M	1	2017	[40]
Yang S	1	2021	[41]
Thamri F	2	2021	[42]
Harputluoğlu N	1	2022	[43]
Perveen S	1	2022	[44]

Literature review/review of the published literature

Below is a table (Table 1) summarizing the published cases in the literature of a Thoracoabdominal duplication. As clarified by this table and supported by the references^[3,10,11,17-44], we concluded that there are only 75 published cases handling this pathology in the literature, making our case the 76th. This conclusion is of profound importance and it exemplifies the magnitude of our findings for the medical community worldwide. The rarity of this disease warrants documentation and conduction of in-depth studies on the subject.

It is worth noting that we did not mark any obstacles during the pre/intraoperative phases. Nevertheless, the primary limitation we encountered was the lack of some of the advanced equipment needed for conducting this type of surgery in a minimally invasive manner.

Conclusion

Thoracoabdominal duplication is a rare congenital pathology, as demonstrated by the lack of the corresponding sufficient data in the literature. Nevertheless, so is intestinal malrotation. These congenital anomalies should be documented to enrich the literature through accurate depiction and illustration of such potentially fatal pathologies. Early detection and timely surgical interventions pave the path for a good prognosis and decrease the subsequent morbidity and mortality.

Based on our experience with this topic, our recommendations are to perform surgical interventions as soon as possible, avoid any negligence or postponing of the therapeutic intervention, and perform the surgical procedure in two steps rather than one (The thoracic part as a first step and the abdominal part as a second step) because this helps decrease the operation time and therefore, limit the morbidity and mortality.

Upon detailed review of the available published literature, we can conclude that ours is the first-ever case documented from our country regarding this topic and this co-occurrence.

Ethics approval and consent to participate

Institutional review board approval is not required for deidentified single case reports or histories based on institutional policies.

Consent of patient

Written informed consent was obtained from the patient's parents/legal guardian for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Author contribution

O.A.: Conceptualization, resources, methodology, data curation, investigation, who wrote, original drafted, edited, visualized, validated, literature reviewed the manuscript, and the corresponding author who submitted the paper for publication.

Q.M.: First assistant during the operation, in addition to resources, data curation, investigation, who wrote, original drafted, edited, visualized, validated, and literature reviewed the manuscript.

H.O.: Data curation, resources, validation, visualization, writing, and reviewing the manuscript.

R.S.: Data curation, resources, validation, visualization, writing, and reviewing the manuscript.

S.A.: Histopathology specialist who analyzed the resected specimens, validated, and reviewed of the manuscript.

M.A.: Pediatric surgery specialist who supervised the operation, in addition to supervision, project administration, and review of the manuscript.

All authors read and approved the final manuscript.

Conflicts of interest disclosure

Not applicable.

Research registration unique identifying number (UIN)

Not applicable.

Guarantor

Omar Al Laham.

Availability of data and materials

The datasets generated during and/or analyzed during the current study are not publicly available because the data were obtained from the hospital computer-based in-house system. Data are available from the corresponding author upon reasonable request.

Provenance and peer review

Not commissioned, externally peer-reviewed.

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