CASE REPORT

Enteric duplication cyst—a peculiar aetiology of anaemia in a two-year- old boy: a case report

Fanzer Khan, Nabiha Eeman, Ahsan Ali Mirza, Atta ul Rehmen Saadi

Abstract

Enteric duplication cysts are rare congenital malformations of gastrointestinal tract that are characterised by the presence of epithelial lining of intestinal mucosa, a layer of smooth muscle and a common wall with the GI tract that may or may not show communication with the gut lumen. On the basis of structural classification, enteric duplication cysts can be either cystic or tubular. This case report is about a two-year-old boy who presented to the Outpatient Department of Paediatrics at the PAEC General Hospital, Islamabad, with complaints of black tarry stools and lethargy since two months and cough since one month. He had a history of multiple blood transfusions. After detailed examination and investigations, diagnosis of thoracic enteric duplication cyst was proposed. Thoracic cystectomy was done. Diverging from its usual respiratory or dysphagic manifestations, this thoracic enteric duplication cyst (extending above into the neck and below into the parahepatic space) presented as a diagnostic enigma through severe anaemia and gastrointestinal bleeding, highlighting its extraordinary rarity.

Keywords: Anaemia, (D000740), Cyst (D003560), Congenital abnormality (D000013)..

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Introduction

Congenital malformations are characterised by structural or functional defects that occur during the foetal development and can be identified prenatally, at birth, or in infancy.[1] Accounting for 15% of all congenital malformations, gastrointestinal malformations include a variety of disorders like atresia, stenosis, intestinal malrotation, duplication, and Hirschsprung's disease.[2]

Enteric duplication cyst is a rare congenital malformation involving the alimentary tract (prevalence is 1 in 4,500 live births with a slight male predominance). It is characterised by the presence of epithelial lining of intestinal mucosa, a layer of smooth muscle, and a common wall with the GI tract (serous or muscle membrane) that may show communication with the lumen of GI tract.[3] The duplication can exist anywhere along the GI tract with majority of cases originating from the mid-gut.[4] Duplication cysts are most commonly located on the mesenteric border of the gut; ileum being the most common site. They are broadly classified into two categories, based on their structural configuration: cystic (type 1:79%) and tubular (type 2:21%).[3] A diverse clinical presentation ranging from intestinal obstruction, intussusception, perforation, bleeding, and chronic intermittent abdominal pain is seen due to variation in location, size, and presence of heterotopic tissue. There is also a risk of malignant transformation specifically in adults.[5] Consent was taken from the parents for recording of data and reporting the findings of the case.

Case Report

A two-year old boy, without any previous medical history, presented to the Outpatient Department of Paediatrics at the PAEC General Hospital, Islamabad, in July 2024 with complaints of black tarry stools and lethargy since two months and cough since one month. He had a history of 2 blood transfusions over the past 2 months. There was no history of bleeding from other sites of the body. No associated family history of bleeding diatheses, genetic bleeding disorders, or liver disease was noted.

On general physical examination, the child was pale. Both height and weight of the child were between 5th and 10th percentile on the growth chart. On chest examination, decreased breath sounds were appreciated on the right side. Abdomen was soft, non-distended and non-tender with normal bowel sounds. The rest of the systemic examination was insignificant.

Baseline investigations were done. Complete Blood Count (CBC) report showed microcytic hypochromic anaemia with haemoglobin 11.1 g/dl (12-14 g/dl), total leukocyte count of 11,960/µL (4500-13,500/µL), and platelet count of 671,000/µL (150,000-450,000/µL). Chest X-ray showed an elongated opacity with air lucency on

Department of General Surgery, Pakistan Atomic Energy Commission General Hospital, Islamabad, Pakistan.

Correspondence: Nabiha Eeman. **Email:** n.eeman4473@gmail.com

ORCID ID: 0009-0008-6351-9949

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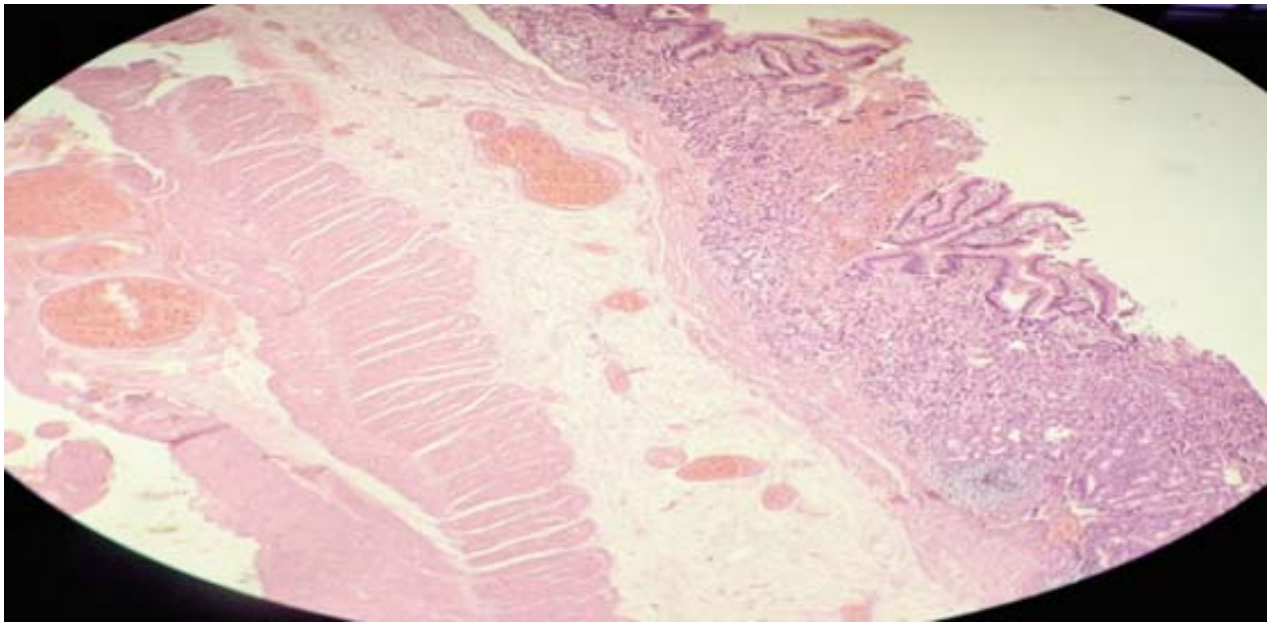

Figure-1: Enteric (gastric) Duplication Cyst: the above specimen reveals a fibro muscular cyst wall lined by simple columnar epithelium with underlying gastric glands. No evidence of dysplasia or malignancy is noted.

the right side of the upper mid and lower mediastinum. CT of the chest and abdomen showed a large, relatively well-defined multiloculated and multiseptated collection with air fluid levels in the right paraspinal region of mediastinum, extending from thoracic inlet up to the para hepatic region traversing the diaphragm with surrounding stranding and oedematous changes with enlarged enhancing mediastinal lymph nodes. Swallow contrast study showed right posterior mediastinal mass with air fluid levels at all stages of the study, but did not show any communication with any gut loops. All these findings strongly suggested the diagnosis of Enteric duplication cyst.

Right thoracotomy was done. Following were the per-operative findings: large enteric duplication cyst (macroscopically identified to be accessory stomach) in the thorax extending up to the neck and below via the diaphragm into the abdomen (in parahepatic space). Cystectomy was performed. The upper end of the cyst was extending into the neck and could not be reached and duplicated gastric fundus had to be left (gastric mucosa was fulgurated by diathermy). Lower extension of the cyst was dissected and diaphragmatic hernia was repaired using Vicryl 2-0. Chest tube was placed in the thorax, haemostasis was secured and closure done.

Post-operatively, the patient was closely monitored. He passed flatus and stool two days later and oral nutrition was introduced gradually. Chest tube was removed after five days. The patient made an uneventful recovery and was discharged after one week. Histopathology report confirmed the diagnosis of enteric duplication cyst with lining of gastric mucosa (accessory stomach) with no dysplastic or malignant changes as shown in Figure 1. The acid secreting nature of this heterotopic gastric tissue within the cyst (present in 20%-30% of the cases) can lead to significant complications such as gastrointestinal bleeding, abdominal pain, obstruction, perforation, and rarely malignant transformation, thus necessitating prompt diagnosis and surgical excision. Presently, the patient is in optimal clinical condition and undergoing semestral follow-up.

Discussion

Gastrointestinal duplication cysts are rare congenital anomalies with an estimated prevalence of 1 in 4,500 live births.[3] According to Ladd, the three essential features that should be present in order to label a cystic lesion as enteric duplication cyst include: 1) smooth muscle coat, 2) epithelial lining resembling GI tract, and 3) close anatomical location with some part of the gut.[6] While enteric cysts can occur anywhere along the alimentary tract, their presentation as a cervicothoracic-abdominal lesion, extending from the neck, through the diaphragm, and into the parahepatic space is exceptionally rare.

Various hypotheses have been proposed in order to explain the process of intestinal duplication. According to Bentley who proposed the split notochord theory, incomplete separation of notochord from gastrointestinal endoderm during the third and fourth week of gestation

results in gaps in the notochord that may cause the gut endoderm to herniate and form diverticula[7]. According to defective recanalisation theory, GI organs begin as solid structures and vacuolate to form lumens. During this process, diverticula form but regress during foetal life. If they persist, duplications could form[8]. The embryonic diverticula theory postulates that an existing diverticula in the growing embryo will persist and grow separately from the alimentary tract[9]. Vascular accident theory states that alimentary tract duplication is a result of focal areas of vascular insufficiency secondary to foetal stress and anoxia.[10] According to abortive twinning theory, organs can be doubled as a result of partial twinning. It is most commonly used to explain cysts of colorectal or genitourinary system.[11]

Thoracoabdominal cysts account for less than 2% of alimentary tract duplications. While gastrointestinal duplication cysts are rare, the extensive multi-compartmental involvement, observed in the current patient, specifically a continuous lesion extending from the cervical region through the thoracic cavity and inferiorly into the parahepatic space, represents an exceptionally uncommon presentation. A comprehensive literature review reveals only a limited number of reported cases describing duplication cysts with significant longitudinal extension or involvement of multiple body compartments. S Moralioğlu et al.[12] presented a case of Thoracoabdominal duplication cyst in an eight-month-old boy with respiratory distress and abdominal pain. Their case involved multiple compartments similar to the current case but the predominant clinical presentation was respiratory distress and not anaemia. A diverse clinical presentation can be seen due to the variation in size, location, and presence of heterotopic tissue. According to Macpherson, ectopic gastric mucosa can be seen in 29% of thoracoabdominal cysts.[13] The presence of ectopic gastric mucosa can lead to gastrointestinal bleeding and anaemia as reported in the index case.

Various investigations are available to diagnose enteric duplication cysts. Plain radiographs aid in the diagnosis of enteric duplication cysts present in the mediastinum. Ultrasound is the most common imaging modality to diagnose intra-abdominal duplications with "classic gut wall signature" that is characterised by the presence of cystic rim of hyperechoic serosa and an inner hyperechoic rim of the mucosa and submucosa with a hypoechoic muscular layer sandwiched between the two hyperechoic layers. CT and MRI can be used to describe the location and size of the cyst which will further help in surgical management. Contrast studies may show a mass effect or communication with the alimentary tract. Endoscopy or laparoscopy can be helpful in the diagnosis of enteric duplication cysts in the absence of significant findings on imaging[14].

The treatment is mainly surgical excision with the aim to remove the duplication but it should not be more radical than necessary to eliminate patient's complaint and prevent recurrence[12]. After surgical excision, histopathological assessment of the specimen is very important to find out about the potential malignant transformation of duplication cysts, which aids in further treatment plan.

Conclusion

This report illustrates a particularly unusual presentation of a thoraco-abdominal enteric duplication cyst in a two-year-old child, manifesting as recurrent melena and severe anemia rather than the more commonly described respiratory or dysphagic symptoms.Its thoraco-abdominal extension made clinical diagnosis challenging, yet targeted imaging enabled accurate identification. This case emphasizes that enteric duplication cysts should be considered in the differential diagnosis of unexplained gastrointestinal bleeding or anemia in children. Early recognition and timely surgical excision can prevent morbidity. Reporting such rare and atypical cases contributes to the medical literature, raising awareness among clinicians and aiding prompt diagnosis and management in future patients with similar presentations.

Consent form: Informed consent was taken from the patient's guardians prior to the submission of the manuscript for publication.

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AUTHOR'S CONTRIBUTION:
FK: Concept, assistance during the clinical intervention and literature review.
NE: Assistance during the clinical intervention, writing original draft, editing and literature review.
AAM: Surgical intervention, responsible for integrity, review and final approval.
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