

Clinicopathological Features of Enteric Duplication Cysts in Children: A Five Year Study

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Received: November 19, 2019; **Published:** January 09, 2020

Abstract

Introduction: Enteric duplication cysts (EDC) are rare congenital defects of gastrointestinal tract. They have wide clinical presentation owing to their location and presence of ectopic tissue or lining.

Materials and Methods: Thirty cases of EDC were retrieved from records in past five years and detailed clinicopathological details were studied.

Results: Age ranged from 4 days to 9 years with 60% cases being infants. Most common presenting complaint was abdominal pain. Commonest site was ileum. Unique histopathological features included gastric, respiratory lining epithelium, ectopic pancreatic rests, segmental absence of intestinal muscle, dystrophic calcification, lymphangiectasia and associated ileal and jejunal atresias.

Conclusion: Histopathology is gold standard in establishing diagnosis in EDC. They can be associated with other congenital GI anomalies. Early diagnosis with appropriate intervention may prevent associated untoward events like torsion, perforation, bleeds from ectopic gastric lining and though rare malignant transformation.

Keywords: Duplication; Cysts; GIT; Enteric Cysts; Ectopic Gastric Mucosa; Ectopic Pancreas

Abbreviations

EDC: Enteric Duplication Cyst; CT: Computed Tomography; USG: Ultrasonography

Introduction

Enteric duplication cysts are rare congenital malformations seen mainly in infants and children with an incidence as low as 0.2% [1,2]. They can be seen anywhere along the alimentary tract from the mouth to anus and may be associated with other congenital anomalies of the GI tract. Midgut duplications are the commonest, followed by foregut and hindgut [3]. The exact embryogenesis of these cysts is yet to be determined, however various theories have been proposed. These lesions pose a diagnostic dilemma to both clinicians and radiologists due to their wide variation in clinical presentation and nonspecific symptoms that depend mainly on their location, type, size and presence of ectopic tissue. Hence, histopathological examination is the gold standard in establishing diagnosis. We present thirty cases of duplication cysts with some unique histopathological features and coexistent uncommon clinical features.

Materials and Methods

This is a retrospective study carried out in the Departments of Pathology and Pediatric Surgery, Chacha Nehru Bal Chikitsalaya, New Delhi, India. Thirty cases of enteric duplication cysts over a period of five years from January 2014 to December 2018 were selected. These cases were partly diagnosed clinico-radiologically and confirmed on histopathology. Data regarding patient age, sex, clinical presentation, radiological features and intra-operative findings was retrieved from the hospital records and analyzed. The gross features and histopathology slides were reviewed for the lining epithelium and other associated histopathological features.

Results and Discussion

Thirty cases of histopathologically confirmed duplication cysts were included in the study. The age of the patients ranged from 4 days to 9 years with eighteen patients (60%) being infants. 10 out of these 18 were neonates at the time of diagnosis. Males were affected more than females with M: F ratio of 4:1.

The commonest clinical presentations were abdominal pain (59%), abdominal distension (53%), palpable lump (35%), vomiting (35%) and non-passage of stools (24%). A single patient with cyst in the mediastinum presented with respiratory distress. The clinical presentation of the cyst was related to the site of the cyst.

Radiological investigations were variable. X-ray was done in all the cases and showed air-fluid levels in five of them. Ultrasound and CT scan were done in eighteen cases. A provisional diagnosis of duplication cyst was given in only twelve cases and in the others; differential diagnosis of mesenteric cyst, infective cyst, pseudocyst and omental cyst were given. Figure 1a shows CT of a child with a relatively well defined lesion with fluid attenuation and few septae. Lesion is occupying the entire right hemithorax with an intrabdominal extension. Figure 1b shows X-Ray of another child showing an opaque right hemithorax with tracheo-mediastinal shift to left suggesting a right sided mass lesion? Cyst.

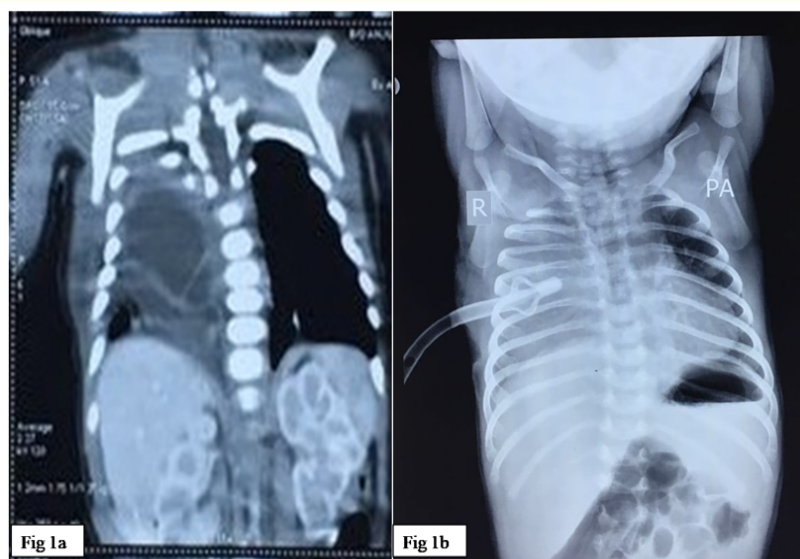


Figure 1: 1a: CT scan showing a relatively well defined lesion with fluid attenuation contents and few septae. Lesion was occupying the entire right hemithorax with an intrabdominal extension. 1b: X-Ray image of a child showing an opaque right hemithorax with tracheo-mediastinal shift to left suggesting a right side mass lesion? Cyst.

The commonest site for duplication cyst was ileum (60%) followed by jejunum (20%), colon (10%), duodenum (6%) and mediastinum (3.3%). They had a smooth external aspect and ranged in size from 1 to 18 cms in maximum dimension. Figure 2 shows gross appearance of the cysts. The cyst contents were mucinous in 8 cases, serous in 6 cases and seromucinous in 4 cases. Other twelve were devoid of contents.

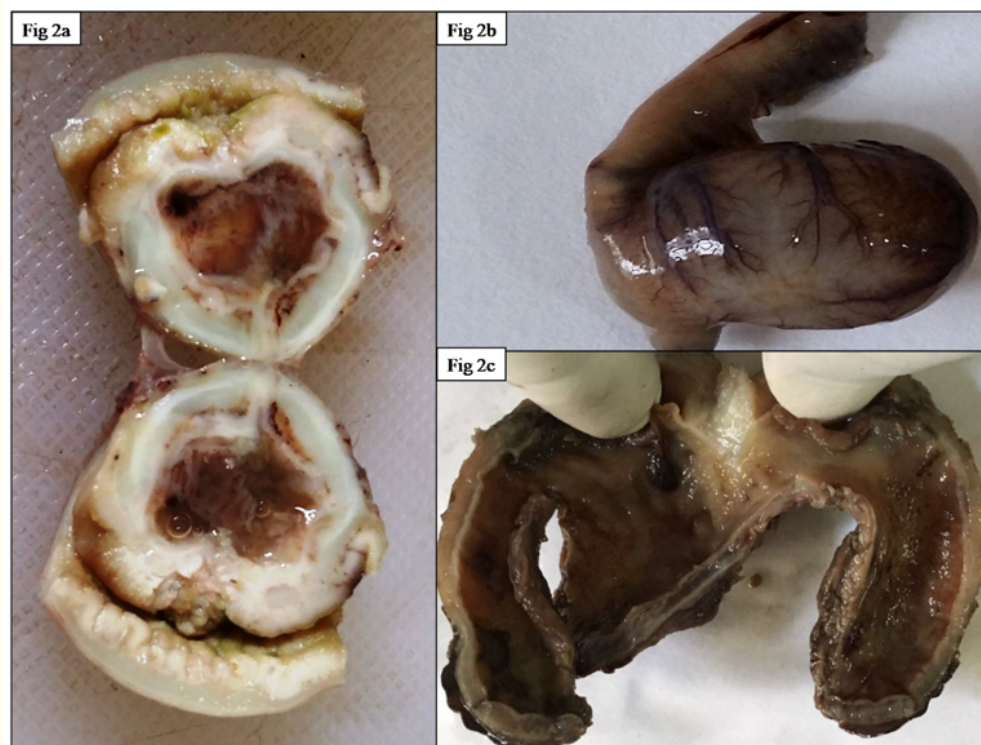


Figure 2: 2a: Gross image of a cyst measuring 3 x 2.5 x 2 cms with adjacent normal appearing intestine. Cyst was filled with serous fluid. 2b: Gross image of a communicating cyst. Externally, the cyst is markedly congested. 2c: Cut section of the same shows an ovoid cyst communicating with the lumen, devoid of contents.

The various histomorphological features of these cases have been highlighted in table 1, which include the variable lining epithelium (Figure 3a and 3b), ulceration with presence of only acute inflammatory granulation tissue, presence of ectopic rests (Figure 3c), dystrophic calcification (Figure 3d). We came across with rare associations like segmental absence of intestinal muscle, lymphangiectasia and intestinal atresia. The other associated features are highlighted in table 1.

Duplications of the alimentary tract are rarely seen congenital malformations with an incidence of 1:4500 [4]. The term intestinal duplication was first used by Fitz in 1844 [5]. These are usually diagnosed in the first year of life and are common in males. In the current study males were affected more than the females with majority being infants. Parker, *et al.* reinforced the criteria laid down by Ladd and Gross for diagnosis of duplication cysts [6,7]. The diagnosis requires presence of a double layered muscle coat with a lining epithelium similar to the gastrointestinal tract and a close association or adherence to any part of the intestinal tract [8]. Also, the duplication cysts are seen commonly along the mesenteric border, share a common blood supply with the bowel wall and show absence of any communication with the lumen of the bowel [9].

S. No	Characteristic histopathological features	No. of cases	Previously reported
1	Intestinal lining	16	Yes
2	Ectopic gastric epithelium	5	Yes
3	Respiratory epithelium	1	Yes
4	Squamous metaplasia	8	Yes
5	Ulcerated lining with dense acute inflammation	9	Yes
6	Foreign body giant cell reaction	5	Yes
7	Dystrophic calcification	6	No
8	Ectopic pancreatic rests	5	Yes
9	Lymphangiectasia in adjacent intestine	2	No
10	Segmental absence of intestinal muscle	1	No

Table 1: Various histopathological features.

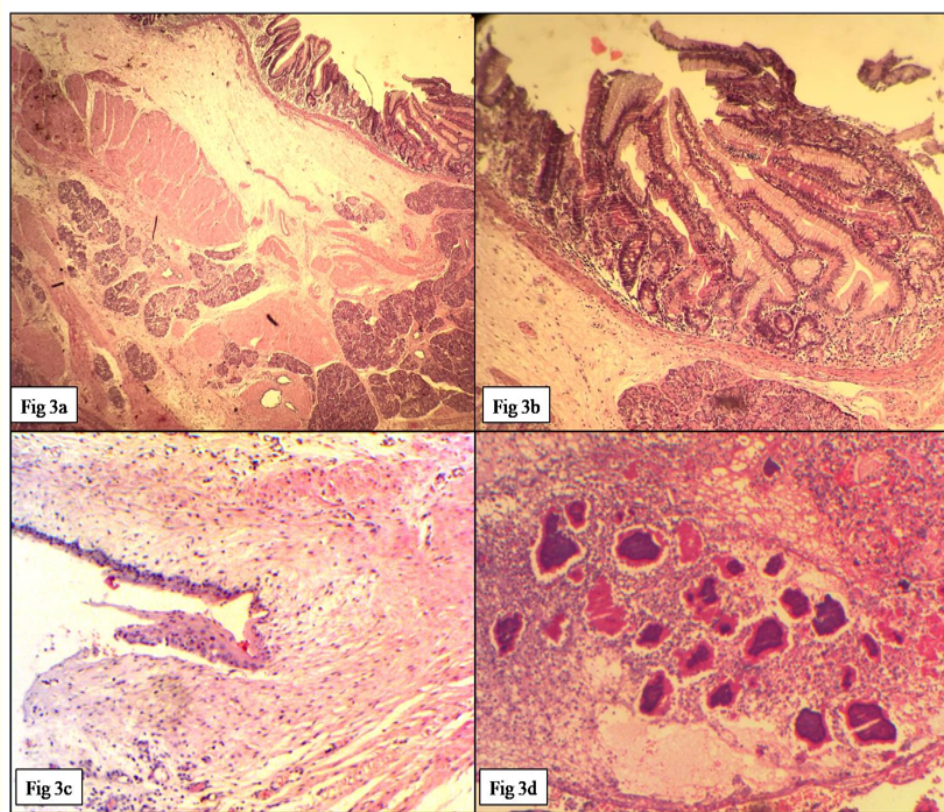


Figure 3: 3a: Photomicrograph of cyst lined by intestinal epithelium with presence of ectopic pancreatic rests in the submucosa. (Hematoxylin and Eosin, 100 X) 3b: Photomicrograph of cyst lined by gastric mucosa. (Hematoxylin and Eosin, 100 X). 3c: Cyst wall lining showing focal squamous metaplasia along with ulcerated lining epithelium. 3d: Cyst wall showing dystrophic calcification.

The embryogenesis of these cysts is a topic of discussion with several proposed theories regarding their origin like split notochord theory, theory of abortive twinning, persistent embryonic diverticula theory, aberrant luminal recanalization theory and intrauterine vascular accident theory [10]. Of these, the most widely accepted is the split notochord theory for the neuroenteric cysts. According to this, the developing notochord and the primitive endoderm of foregut fail to separate, leading to the persistence of neuroenteric canal and formation of the neuroenteric cyst - these duplication cysts are associated with vertebral anomalies. In the present study none of the cases had associated vertebral anomalies.

The abortive twinning theory proposes that gastrointestinal tract duplications represent incomplete twinning. The persistent embryonic diverticula theory suggests that the transient diverticula on the antimesenteric aspect of the intestinal wall of embryos persist and develops into duplications. The theory of aberrant luminal recanalization explains duplications in parts of alimentary tract that go through the solid stage such as esophagus, small bowel and colon. The intrauterine vascular accident theory suggests that gastrointestinal tract duplications arise as the result of intrauterine vascular accidents. All of these theories however fail to explain the origin of the duplication cyst completely [10].

Cysts are named depending upon the part of the alimentary tract with which the cyst is associated, the commonest being ileal enteric duplication cyst [1,11]. In the present study too ileum was the commonest site. Colon, rectum, stomach and mediastinum are rarer sites for duplication. The clinical presentation varies, depending on the location of the cyst, their size and presence of ectopic tissue. They usually present in childhood and the usual clinical presentations are abdominal pain, distension, palpable lump, vomiting and non-passage of stool [12]. Some of the cysts remain asymptomatic and present very late in life. Large cysts in the mediastinum may cause respiratory distress due to pressure effect [13]. Gastric epithelium in the cyst wall may ulcerate and bleed or even perforate leading to peritonitis. If left untreated, malignant transformation of gastric epithelium may be seen [14]. Ectopic pancreatic rests, when present may lead increased pancreatic enzymes within the cyst fluid and also to hypoglycemic attacks which are very difficult to diagnose in children.

The presumptive diagnosis of these cysts can be given by their clinical presentation and radiological features. The various imaging modalities used are X-rays, barium studies, ultrasonography (USG) and computed tomography (CT) scan [15]. They are likely to suggest the diagnosis in majority of cases. A plain abdominal X-ray may help detect a soft tissue mass. Barium studies reveal an intraluminal, intramural or extrinsic mass related to the alimentary canal. USG is the imaging modality of choice and demonstrates the location of the mass and its cystic nature. It shows presence of inner echogenic mucosal layer and adjacent hypoechoic muscle layers within the cyst. CT scan demonstrates the exact location and extent of the lesion with presence of associated vertebral anomalies if present. Most of the duplication cysts appear as smoothly rounded, fluid filled structures with slightly enhancing wall adjacent to intestinal tract. In the present study a radiological definitive diagnosis of duplication cyst could be given in twelve out of thirty cases. In majority cases, diagnosis was established on USG and CT. Tc-99m pertechnetate scintigraphy can also be performed to demonstrate the presence of ectopic gastric mucosa within the cyst wall.

Histopathological examination is the gold standard in diagnosis of enteric duplication cysts. In the present study histopathology confirmed the diagnosis in all the cases. All the cases had double layered muscle wall and were seen in close association to some part of the gastrointestinal tract with presence of the lining epithelium in majority of the cases. None of the cysts were communicating with the intestinal lumen. The characteristic histopathological features and other associations are highlighted in table 1. Ectopic pancreatic rests rarely reported in literature, was seen in five cases [16]. A single case had coexisting segmental absence of intestinal muscle with EDC, an entity not reported in Literature earlier. EDC can be associated with intestinal atresia as reported by Sinha., *et al* [17]. This association can be explained by the vascular accident theory of the origin of these cysts. In the present study we had two cases of duplication cyst associated with ileal and jejunal atresia. Xiao Ming., *et al.* reported one case of EDC with three different lining epitheliums within the same cyst; we found only one case with coexistent respiratory and intestinal lining [18].

The differential diagnosis of EDC includes mesenteric cyst, omental cyst, pancreatic pseudocyst, choledochal cyst, volvulus and intussusception [11,19]. These can be differentiated on the basis of their clinico-radiological findings and histopathological features. Mesenteric and omental cysts are endothelium lined cysts which lack the double layered muscle wall of duplication cysts. Pancreatic pseudocyst is seen in or near the pancreas, has a fibrous wall and no epithelial lining. Choledochal cysts are cystic dilatations of the biliary tree and have columnar epithelium lined fibrous wall. In volvulus, there is torsion of the bowel loop leading to ischemia and gangrene of the affected segment of bowel. Histopathology reveals transmural gangrene of the bowel wall. In intussusception one portion of the bowel invaginates into the adjacent portion leading to symptoms of obstruction. Usually, there is a lead point which leads to intussusception. Histopathology reveals gangrenous or ischemic changes depending on the duration and amount of occlusion of blood supply.

Outcome of a cyst without associated anomalies is favourable. The treatment of choice is surgical excision [20]. Asymptomatic cysts need to be removed to avoid late complications like torsion and rarely malignant change. Symptomatic EDC require urgent intervention. The cyst is excised along with adherent intestine and end to end anastomosis is done. The postoperative period is usually uneventful. In the present study all the patients recovered without any complications and are doing well at follow up.

Conclusion

Enteric duplication cysts are an uncommon form of congenital defect in children and a high index of suspicion is required for their diagnosis, both clinically and radiologically due to its wide clinical presentation. Ileum is the commonest site. They can be associated with several other congenital gastrointestinal anomalies. Histopathology is gold standard and can show variable features. An early pre operative diagnosis with quick intervention is imperative in this easily treatable condition to ensure better outcome. To date, it is the largest series of duplication cysts reported with such variable histopathological features.

Conflict of Interest

None.

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Volume 7 Issue 2 February 2020

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