Original Article

DOI: 10.7860/JCDR/2015/12929.6381

Enteric Duplication Cysts in Children: A Clinicopathological Dilemma

Pathology Section

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ABSTRACT

Aim: Enteric duplication cysts are rare and uncommon congenital malformations formed during the embryonic period of the development of human digestive system and are mainly encountered during infancy or early childhood, but seldom in adults. The clinical presentation is extremely variable depending upon its size, location and type. We present six cases of enteric duplication cysts with diverse clinico-pathological features.

Materials and Methods: This study was carried out in the Department of Pathology and Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, India for a period of 2 years (January 2013 - December 2014). We retrospectively analyzed six patients of enteric duplication cysts based on data obtained, which consisted of patient's age, sex, clinical presentation, radiological features, operative findings and histopathology report. The data collected was analyzed by descriptive statistics.

Results: Six children between age range of 3 days to 10 years had enteric duplication cysts. Two had ileal and one each were of pyloroduodenal, colonic and rectal duplication cyst. In one patient a presumptive diagnosis of enteric duplication cyst was made. Radiology played an important contributory role in diagnosis of these cysts in all the patients but histopathology proved to be gold standard for its confirmation. All these patients were managed by surgical excision. The postoperative and follow up period in all the cases was uneventful.

Conclusion: It is important to be aware and make a definitive diagnosis of this rare congenital anomaly as they can present in various clinical forms and can cause significant morbidity and even mortality if left untreated by causing life threatening complications.

Keywords: Congenital anomaly, Histopathology, Surgical excision

INTRODUCTION

Enteric duplication cysts are rare developmental anomalies, originating anywhere along the alimentary tract from the tongue to the anus. The reported incidence is 1:4,500 births [1]. Most duplications are detected in children (antenatally or within first two years of life) and fewer than 30% of all duplications are diagnosed in adults [2,3]. They pose a diagnostic dilemma and therapeutic challenge, as the presenting symptoms are non-specific and variable which are closely related to the type, size and location of the cyst. Approximately 75% of duplications have been reported to be located within the abdominal cavity, whereas the remaining is intrathoracic (20%) or thoracoabdominal (5%). Ileal lesions are the most common (53%), followed by mediastinal (18%), colonic (13%), gastric (7%), duodenal (6%), rectal (4%), oesophageal (2%) and cervical (1%) lesions. They may present as solid or cystic tumours, intussusception, perforation, or gastrointestinal bleeding. Hence, high index of suspicion is required in such cases. Enteric duplication cysts may be an incidental finding intraoperatively but in the majority of cases preoperative radiological diagnosis is possible. Appropriate surgical management is required, for this the attending surgeon should be familiar with the pathology and clinical characteristics of these rare cysts.

Due to the rarity of this condition the vast majority of literature on enteric duplication cysts is in the form of case reports. Very few case series have been published previously [4,5]. On extensive PubMed and Medline search, it was found that no case series has been published from India.

MATERIALS AND METHODS

This was a retrospective study carried out in the Department of Pathology and Department of Paediatric Surgery, Vardhman Mahavir Medical College and Safdarjung Hospital, New Delhi, India. All patients who were clinically diagnosed and proved on histopathological examination to be of enteric duplication cysts were included in the study. For the purpose of analysis information was obtained from patient medical records and histopathology records. The data that was gathered consisted of patient's age, sex, clinical presentation, radiological features, operative findings and histopathology report. The data collected was analysed by descriptive statistics.

RESULTS

There were a total of six patients during the study period of two years (January 2013 - December 2014). There were 4 males and 2 females (Male: Female = 2:1). The age of patients ranged from 3 days to 10 years. Four patients (66.7%) were less than 1 year old. Out of these, three (75%) patients presented during first 10 days of life.

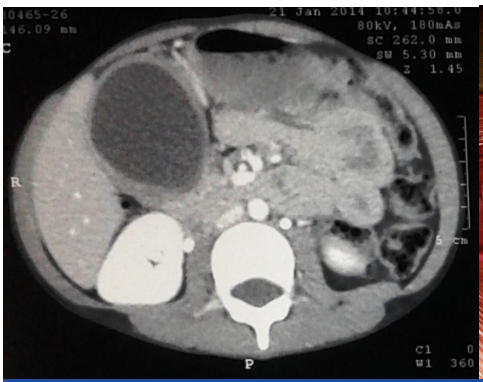
[Table/Fig-1] summarizes the age, sex, clinical presentation, physical examination, radiological investigations, operative findings, gross, microscopic examination and final diagnosis of all the six patients. Ultrasound (USG) and computed tomography (CT) were able to identify the cystic masses in all six cases. Enteric duplication cyst was the primary diagnosis in all patients except patient no. 5 in which it was part of the differential diagnosis.

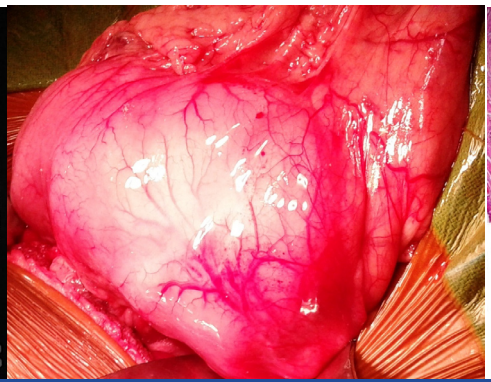
Histopathological examination showed presence of mucoid material in four (66.6%) cases. In two cases (33.4%) no cyst contents were seen. The size of the cysts varied from 2 cm to 25 cm. Lining epithelium was identified in five cases (83.3%). No ectopic tissue could be identified in any of the cases.

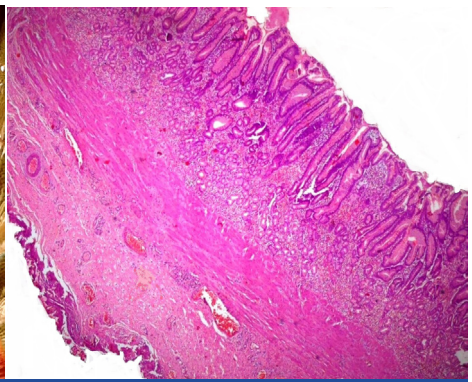
Out of the six patients two (33.4%) had ileal duplication cysts. There was one patient each of pyloroduodenal, colonic and rectal duplication cyst [Table/Fig-2-8]. In one patient a presumptive diagnosis of enteric duplication cyst was made because no lining epithelium was seen despite extensive sampling. However, the facts that the cyst was in close relation to bowel loops and had smooth muscle in cyst wall arranged in circular and longitudinal manner strongly favoured a diagnosis of enteric duplication cyst.

Case	Age/Sex	Clinical presentation	Physical examination	Radiological investigations	Operative findings	Surgical procedure	Gross	Microscopic examination	Final diagnosis
1	6 yr/ Female	Abdominal pain, abdominal lump and infrequent episodes of non bilious projectile vomiting	Vague mass palpable in right upper abdomen, tenderness on deep palpation	USG abdomen - well defined uniloculated cystic lesion in the pyloro-duodenal region measuring 3 × 3 cm. CT scan abdomen - cystic mass measuring 3.5 × 3 cm at the pyloro-duodenal junction [Table/Fig-2]	Cystic mass measuring 4 × 3 cm at the pyloro-duodenal junction [Table/Fig-3]	Cyst excision with end to end anastomosis	Cyst measuring 4x3 cm. Lumen contained mucoid material.	Cyst wall lined by gastric mucosa. Wall of the cyst showed smooth muscle. [Table/Fig-4]	Pyloro-duodenal duplication cyst
2	4 day/ Male	Distended abdomen since birth and failure to pass meconium	Firm mass palpable in the right upper quadrant	X-ray abdomen - large air filled structure in the right upper quadrant. USG abdomen - large cystic mass adherent to the hepatic flexure measuring 6x5 cm.	Large cystic mass of sigmoid colon adherent to right upper quadrant. The cyst was not communicating with the gut.	Excision of the cyst along with excision of the segment of sigmoid colon and stapled colocolostomy	Thick walled cystic mass attached to sigmoid colon measuring 6 x 5 cms and filled with mucoid material [Table/Fig-5]	Colonic duplication cyst containing large intestinal mucosa, submucosa and smooth muscle without any ectopic tissue. [Table/Fig-6]	Colonic duplication cyst
3	1 month/ Male	Bilious vomiting and abdominal distention	Cystic firm mass was felt in right lumbar region and bowel sounds were exaggerated	X-ray abdomen- soft tissue density lesion in right lumbar region deviating bowel loops to the left side. USG & CT abdomen – well-defined cystic mass in right lumbar region which was adherent to the ileum measuring 20×5 cm.	Intestinal duplication cyst adherent to ileum measuring 25x5 cm.	Excision of the cyst along with excision of the ileal segment with ileo-ileal anastomosis	Cystic mass with straw coloured mucoid content measuring 25x5 cms. [Table/Fig-7]	Cyst wall lined by ileal mucosa. The muscular layer in the wall of cyst was shared by the cyst and adherent ileum. [Table/Fig-8]	Ileal duplication cyst
4	5 day/ Male	Abdominal distention and failure to pass meconium since birth.	Vague, ill-defined mass palpable in lower abdomen.	USG abdomen - cystic mass in relation to the rectum measuring 2x1 cms.	Cystic mass in close relation to rectum measuring 2x1 cms.	Excision of the cystic mass.	Cyst measuring 2x1x1 cms filled with mucoid material.	Cyst wall lined by rectal mucosa	Rectal duplication cyst
5	10 year/ Male	Abdominal pain, jaundice and abdominal mass	Vague mass palpable in central part of abdomen. Mass was non tender.	CT abdomen - cystic mass in retroperitoneum measuring 6.5x5 cms	Cystic mass present in close relation with bowel loops but not adherent measuring 7x5 cms.	Excision of the cystic mass.	Cyst measuring 7x5.5 cms. No contents seen.	No epithelial lining seen. Wall contained smooth muscle arranged in circular and longitudinal fashion.	? Enteric duplication cyst
6	3 day/ Female	Abdominal distention since birth.	Cystic mass palpable in right mid-abdomen.	USG abdomen - cystic mass adherent to the ileum measuring 4×3 cm. Abdominal CT - uniloculated cyst measuring 5x3 cms.	Intestinal duplication cyst adherent to ileum measuring 4.5x3 cm.	Excision of the cyst along with excision of the ileal segment with ileo-ileal anastomosis	Ileal segment measuring 20 cms in length with attached cyst measuring 4.5x3.5x3 cms. No contents seen.	Cyst lined by ileal mucosa.	Ileal duplication cyst

[Table/Fig-1]: Clinicopathological spectrum of patients with enteric duplication cysts.







[Table/Fig-2]: Pyloroduodenal duplication cyst viewed on CT scan. **[Table/Fig-3]:** Intraoperative pyloroduodenal duplication cyst. **[Table/Fig-4]:** Histology of the pyloroduodenal duplication cyst wall showing mucosa, submucosa and smooth muscle. (H&E,10x)

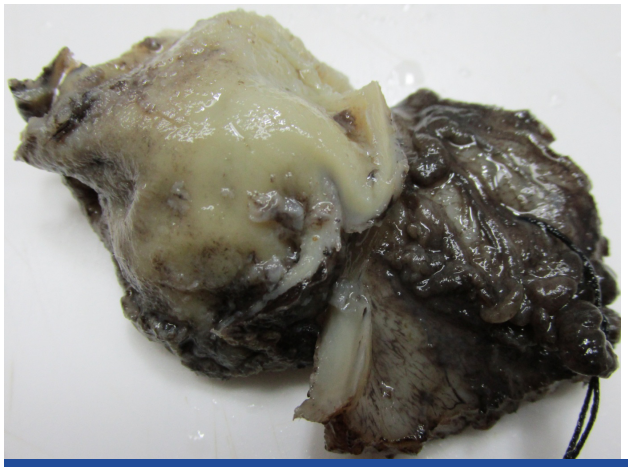
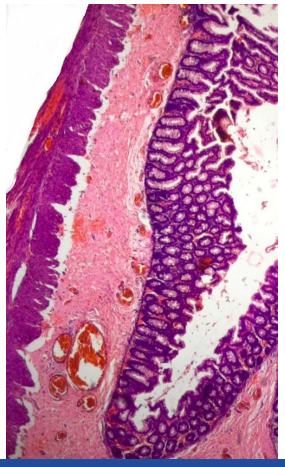
All these patients were managed by surgical excision. In some cases resection of the adherent bowel was done followed by end to end anastomosis. The postoperative period in all the cases was uneventful. On follow up all these patients were found to be healthy.

DISCUSSION

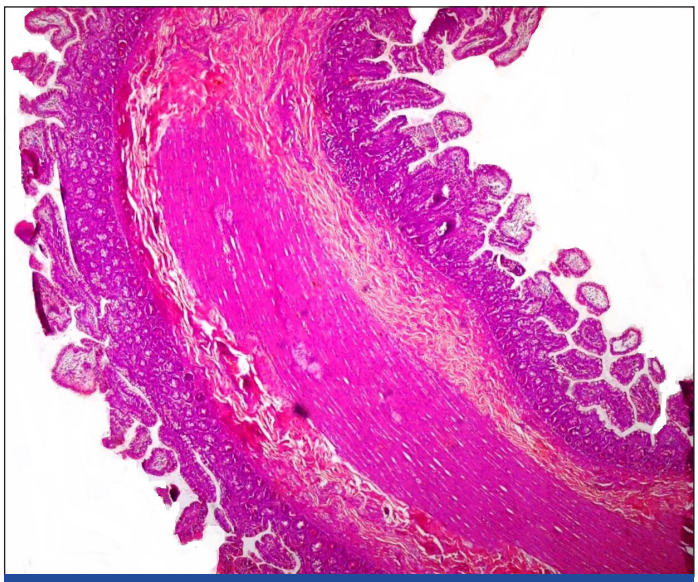
Duplication of gastrointestinal tract is a rare congenital anomaly found in about 0.2% of all children [6,7]. Although enteric duplication cysts can present at any age the vast majority of patients present during infancy. In the present study also four patients (66.7%) were infants.

The term intestinal duplication was first used by Fitz [8] but was not widely used until it was popularized by Ladd in the 1930s,[9] with further classifications by Gross in the 1950s [10]. For the diagnosis of an enteric duplication cyst, three essential characteristics are: (1) a well-developed smooth muscle coat; (2) mucosal lining found within some portion of the alimentary tract; (3) contiguity to any segment of the alimentary tract [11].

Although the exact aetiology is unknown, several theories have been proposed like split notochord (currently most favoured), partial twinning, persistent embryological diverticula or aberrant luminal re-canalization [12]. Other rare causes included intrauterine trauma or hypoxia.


[Table/Fig-5]: Gross specimen of cystic mass adherent to sigmoid colon which is filled with mucoid material. **[Table/Fig-6]:** Colonic duplication cyst wall on histopathology. (H&E,10x). **[Table/Fig-7]:** Gross specimen of excised ileal duplication cyst.



[Table/Fig-8]: Histology of the ileal duplication cyst showing two mucosal layers sharing a submucosal layer with muscular layer. (H&E,10x)

The duplications are named after the part of alimentary tract, to which these are intimately attached. Rai et al., in their series have reported enteric tract duplications with ectopic mucosa like pancreatic tissue [13]. However, in the present study none of the cases showed ectopic tissue. In a large series reported by Pulgandla et al., the most common site for duplications was ileum, accounting for over 60% of cases [4]. Duplication of the colon was found to be quite rare (6.8%) in their series. Murthy [6] and Patel [14] along with co-workers reported, pyloroduodenal duplication cysts to be extremely rare representing just 2.2% of all gastric duplications. In the present study also ileum was found to be the most common site (33.4%). Colonic, rectal and pyloroduodenal cysts which are reported to be extremely rare were also encountered by us in our study.

Many authors have reported variable clinical picture depending on location, size and other factors like presence of ectopic mucosa, communication with adjacent bowel, or inflammation [3]. Infants and neonates present with abdominal pain, nausea, vomiting, bleeding, abdominal distension, abdominal mass, obstruction, intussusceptions [15]. Some duplication cysts may remain asymptomatic till adulthood. In the present study, four of the patients presented with abdominal distension, two with abdominal mass along with other features of intestinal obstruction.

The differential diagnosis includes all causes of neonatal bowel obstruction including volvulus, intussusceptions, mesenteric or omental cysts, a pancreatic pseudocyst, choledochal cyst and infantile hypertrophic pyloric stenosis [15,16]. In this study in one of the patients (patient no.1) infantile hypertrophic pyloric stenosis and in another (patient no. 5) mesenteric or choledochal cyst were considered as differential diagnosis. However, they were ruled out on the basis of clinicoradiological findings and histopathological examination.

The preoperative diagnosis duplication of cysts are often inaccurate. Diagnosis is usually done using imaging modalities such as barium studies, USG or CT scans. Upper gastrointestinal study and barium enema demonstrate filling defect or rarely a communication between the cyst and normal bowel [17]. USG is the imaging modality of choice for the evaluation of an abdominal mass in the neonate which can demonstrate nature and location of the mass. Using scans during pregnancy result in a higher rate of antenatal detection of duplications (approximately 30%) which allow early treatment and avoidance of possible complications [3]. CT scans are more useful in demonstrating the precise anatomical relationship between the cysts and surrounding structures [18]. These cysts can manifest as smooth, rounded, fluid filled cysts or tubular structure with thin slightly enhancing wall on CT scan.

Magnetic resonance imaging (MRI) and endoscopic ultrasonography are other diagnostic modalities. MRI scan shows intracystic fluid with heterogenous signal density on TI weighted image and homogenous high signal intensity on T2 weighted image [19]. Radioisotope scanning is useful for evaluation of bleeding from these cysts. However, all these modalities allow us only to suspect the presence of an abnormal lesion and diagnostic confirmation is possible only after resection. In the present study, USG and CT scan were able to identify the cystic masses in all six cases. Enteric duplication cyst was the primary diagnosis in all patients except patient no. 5 in which it was part of differential diagnosis.

Histopathological examination enables us to confirm the diagnosis. In the present study also, histopathological examination played an important role by helping us to confirm the diagnosis in all the cases. Lining mucosa was identifiable in all cases except one case in which the diagnosis of enteric duplication cyst was presumptive.

Intestinal duplications often require urgent surgical intervention as secretion from ectopic gastric mucosa may lead to ulceration of the intestinal mucosa, massive bleeding or bowel perforation. Treatment of asymptomatic cases remains controversial, though they have to be removed to avoid late complications like malignant change. Small cystic or short tubular duplication are treated by segmental resection along with adjacent intestine. A long tubular duplication is managed by mucosal stripping through a series of multiple incisions, as its excision may lead to short bowel syndrome [20]. In the present study all the patients were managed by surgical resection. In some cases resection of the adherent bowel was done followed by end to end anastomosis. The postoperative recovery and follow up period was uneventful.

CONCLUSION

It is important to be aware and make a definitive diagnosis of this rare congenital anomaly as they can present in various clinical forms and can cause significant morbidity and even mortality if left untreated by causing perforation, obstruction, haemorrhage and malignancy.

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FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Jan 09, 2015**
Date of Peer Review: **May 09, 2015**
Date of Acceptance: **Jun 12, 2015**
Date of Publishing: **Aug 01, 2015**