

## Original Research Article

# Gastrointestinal duplication cysts: experience from a tertiary care center in North India and review of literature

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**Received:** 05 October 2020

**Revised:** 06 December 2020

**Accepted:** 08 December 2020

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## ABSTRACT

**Background:** Duplication cysts (DC) are uncommon congenital malformations which predominantly present in infants and young children. Owing to their variable clinical presentation, radiology and/or histopathology are often required to clinch the diagnosis. We present a case series of 66 patients, which is the largest series reported so far.

**Methods:** A search of prospectively maintained institutional database was carried out to identify patients who were diagnosed and operated for gastrointestinal DC between January 2013 and August 2018. For all cases, the demographic data, site of DC, details of clinical presentation, associated conditions and histopathology findings were recorded. The slides of all cases were retrieved and re-examined.

**Results:** The age range was 1 day to 47 years, with a slight male predominance (1.3:1). Ileum was the most common site followed by jejunum. Ectopic mucosae noted were gastric, pancreatic, biliary and respiratory. One case showed glial heterotopia. Perforation, gangrene and intussusception were among the co-existing pathologies noted.

**Conclusions:** Duplication cysts are rare congenital malformations. The variability in clinical presentation makes the diagnosis elusive. Timely diagnosis and appropriate management require a high index of suspicion and a holistic diagnostic approach with clinical, radiological and histopathological inputs.

**Keywords:** Duplication cysts, Gastrointestinal cysts, Congenital cysts, Gastrointestinal duplication cysts

## INTRODUCTION

Duplication cysts (DC) are uncommon malformations of the gastrointestinal system (GIT) with an estimated prevalence of 1:4500 to 1:10000 in general population.<sup>1</sup> These are cystic structures which, by definition, are located within or adjacent to the wall of a part of GIT, share a layer of their wall as well as their blood supply with the adjacent portion of GIT and have a gastrointestinal mucosal lining same as or different from the adjacent segment.<sup>2,3</sup> They can involve any segment of GIT from the tongue to the anus, however ileum is the commonest reported site followed by esophagus and colon.<sup>2</sup>

The term duplication cyst was introduced by Ladd in 1934 and since then a few case reports and case series have been published from various parts of the world.<sup>4</sup> Duplication cysts are classified into communicating and non-communicating depending on their relationship with the gut. They can also be classified into spherical and tubular based on their appearance. Spherical DCs are the commoner of the two and are usually non-communicating while tubular cysts are usually of communicating type.<sup>4,5</sup> Owing to their rarity, there is limited data on their clinical profile and pathological characteristics. Here we present our experience on duplication cysts from a tertiary care center.

## METHODS

Institutional (Postgraduate Institute of Medical Education and Research, Chandigarh) histopathology database was searched for all cases who were diagnosed and operated for duplication cysts between January 2013 to August 2018. All such cases were included in the study and no cases were excluded. No selection criteria were used, as all cases were included. For all cases, the demographic data, site of DC, details of clinical presentation, associated conditions and histopathology findings were recorded. The slides of all cases were retrieved and assessed for the presence of metaplasia, heterotopia and co-existing pathology, if any. Since it was a retrospective study, no additional consent was required from the study subjects as the consent obtained at time of surgery is inclusive for histopathological analysis. Statistical analyses were

performed with Statistical package for social sciences (SPSS) version 20 (SPSS, Chicago, Illinois, USA).

## RESULTS

A total of 66 patients were diagnosed as duplication cysts in the operated specimens within the study period. Age range was 1 day to 47 years, 54 patients were <12 years of age (our institutional cut off for pediatric patients) and the remaining 12 were >12 years old. Mean age was  $6.72 \pm 11.72$  years. Median age was 1 year. There was male predominance (37 males and 29 females) with male: female ratio of 1.3:1. Most common site of involvement was ileum, followed by jejunum and colon (Table 1).

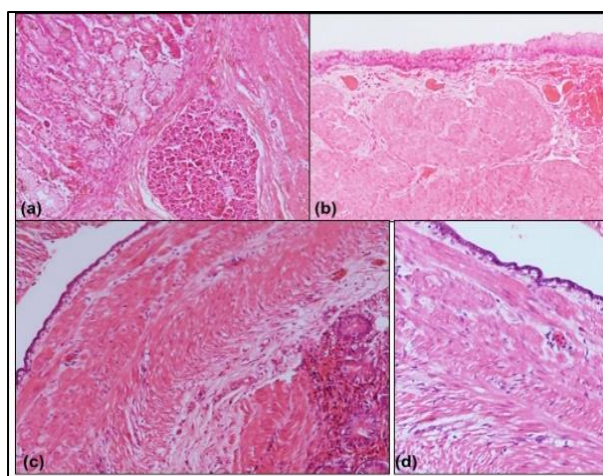
**Table 1: Depiction of the distribution of patients, their demographic characteristics, clinical profiles and pathologic findings according to the site of duplication cyst.**

| Site               | Number | Age                  | Gender                  | Metaplasia                          | Associated findings  | Other clinical findings  |
|--------------------|--------|----------------------|-------------------------|-------------------------------------|--|--|
| <b>Oral cavity</b> | 1      | 31 years             | Female                  | None                                | None   | Sublingual swelling  |
| <b>Esophagus</b>   | 4      | 2 years to 47 years  | Males-3<br>Female-1     | Respiratory-2<br>None-2             | Hemosiderin laden macrophages-1<br>None-3  | Chest pain-1<br>Vomiting-1<br>Mass lesion-2  |
| <b>Stomach</b>     | 4      | 1 day to 10 years    | Males-2<br>Females-2    | Intestinal-2<br>None-2              | Ulceration-1<br>None-3   | Abdominal pain-3<br>Hematemesis-1  |
| <b>Duodenum</b>    | 2      | 7 years              | Male-1<br>Female-1      | Gastric -1<br>Pancreatic-1          | None   | Pain abdomen with vomiting-1<br>Pain abdomen-1   |
| <b>Ileum</b>       | 36     | 1 day to 39 years    | Males- 24<br>Females-12 | Gastric-13<br>Biliary-1<br>None-22  | Ileal atresia-2<br>Calcification-2<br>Gangrene- 5<br>Perforation- 4<br>Ulcer-3<br>Intussusception-1<br>None-19 | Antenatally diagnosed- 2<br>Perforation-4<br>Intussusception-1<br>Acute abdomen-6<br>Pain abdomen-17<br>Intestinal obstruction-6 |
| <b>Jejunum</b>     | 8      | 2 days to 7 years    | Males-4<br>Females-4    | Gastric -2<br>None-6                | Jejunal atresia-3<br>Ulceration-1<br>Congenital diaphragmatic hernia-1<br>None-3                               | Pain abdomen-4<br>Acute abdomen-4  |
| <b>Caecum</b>      | 1      | 3 years              | Female                  | None                                | None   | Pain abdomen   |
| <b>Colon</b>       | 6      | 5 months to 42 years | Male-1<br>Females-4     | Gastric-1<br>Pancreatic-1<br>None-4 | None   | Pain abdomen-3<br>Incidental-2   |
| <b>Rectum</b>      | 4      | 1 month to 14 years  | Males-2<br>Females-2    | None-4<br>(glial heterotopia in 1)  | Ulceration-2<br>None-2   | Esophageal atresia with burst abdomen-1<br>Incidental-1<br>Pain abdomen-2  |

**Table 2: Distribution of cases based on type of mucosal lining of duplication cyst.**

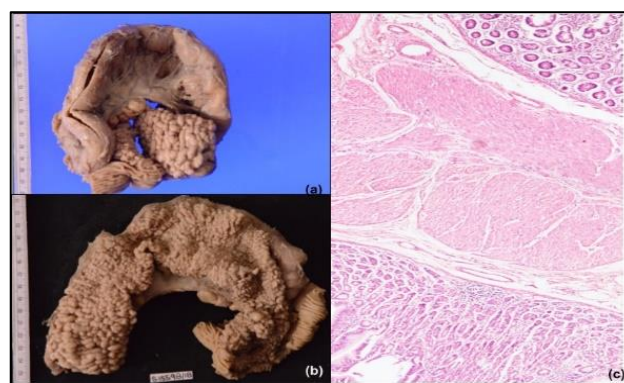
| Mucosal lining   | Cases with mucosa of DC same as that of adjacent segment | Cases with mucosa of DC different from adjacent segment | Total number of cases |
|------------------|--|---|-----------------------|
| Oral cavity      | 1  | -   | 1                     |
| Esophageal       | 2  | -   | 2                     |
| Gastric          | 2  | 17  | 19                    |
| Small intestinal | 28   | 2   | 30                    |
| Large intestinal | 9  | -   | 9                     |
| Biliary          | -  | 1   | 1                     |
| Respiratory      | -  | 2   | 2                     |
| Pancreatic       | -  | 2   | 2                     |
| Total            | 42   | 24  | 66                    |

Respiratory metaplasia was noted in two out of four patients with esophageal DC (Figure 1). Gastric metaplasia was the most common metaplasia, noted in 16 cases with small intestinal (Figure 2) and one case with colonic DC. Biliary type lining epithelium was seen in one case with ileal DC (Figure 1). Three cases showed pancreatic heterotopia, one each with duodenal, colonic and gastric DC (Figure 1). In one patient with rectal DC, there was heterotopic glial tissue in the wall of the cyst (Figure 3) and this one month old male child also had esophageal atresia and presented with burst abdomen. Distribution of the cases based on the type of lining epithelium is depicted in Table 2.

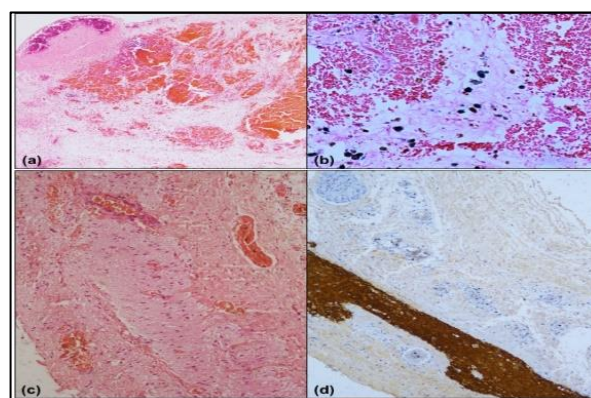


**Figure 1: (a) Enteric DC with pancreatic heterotopia (Hematoxylin and eosin, 100×), (b) Respiratory metaplasia in an esophageal DC (Hematoxylin and eosin, 100×), (c) and (d) Biliary lining in a enteric DC (Hematoxylin and eosin, 100× and 200×, respectively).**

Communicating type of DC (Figure 4) was observed in 32% (21/66) cases (Figure 2). Associated pathologic features were - ulceration in 6 and perforation in 2 cases. Two cases showed extensive calcifications within the cyst wall, along with presence of hemosiderin laden macrophages in one (2-day-old male with ileal DC) (Figure-3) and fibrosis in another (26-day old male with ileal DC).



**Figure 2: Image depicting a non-communicating duplication cyst with extensive gastric metaplasia. (a) A segment of small intestine measuring 35 cm in length with attached duplicated segment throughout its length. The two segments had a common muscularis propria layer. (b) The duplication cyst shows gastric rugae. (c) Representative microphotograph depicting gastric metaplasia in an enteric DC (Hematoxylin and eosin, 40×).**



**Figure 3: (a) Calcification and hemorrhage within the wall of a DC (Hematoxylin and eosin, 100×), (b) Perl's stain highlighting the hemosiderin laden macrophages within areas of hemorrhage (Hematoxylin and eosin, 200×), (c) Glial heterotopia in the wall of rectal DC (Hematoxylin and eosin, 200×), (d) GFAP immunostain highlighting the heterotopic glial tissue within rectal DC (Hematoxylin and eosin, 200×).**



Segment of the DC wall showed gangrene in 5 cases, one of which had associated ileal atresia (2-day-old female with ileal DC). Another 3 day old male patient also had associated ileal atresia. Three cases with jejunal DC had an associated jejunal atresia. A 4-year-old female presented with intussusception and was found to have ileal duplication cyst with biliary metaplasia (Figure 1).



**Figure 4: Image depicting a communicating duplication cyst.**

A segment of small intestine is seen measuring 20 cm in length. 3 cm from one end, a duplicated segment is seen with lumen communicating with the lumen of small bowel.

Pain abdomen and vomiting were the most common presenting symptoms, other symptoms included hematemesis (gastric DC with intestinal metaplasia), bleeding per rectum (ileal DC with gastric metaplasia) and burst abdomen. One of our cases, a 47-year-old male presented with chest pain and after excluding a cardiac event, was found to have an esophageal DC. Two neonates were operated for antenatally diagnosed DC, both of which were ileal in location.

## DISCUSSION

Duplication cysts, which are uncommon congenital malformations of GIT, are identified more commonly in children than in adults.<sup>5</sup> Gastrointestinal duplication cysts may occur within or adjacent to any segment of GIT extending from tongue to anal canal.<sup>2</sup> Owing to their rarity, little data is available on their demographic, clinical and pathological characteristics. It usually remains an unsuspected diagnosis clinically and is commonly detected radiologically. Otherwise, the diagnosis tends to be established during the course of the histopathologic examination of the resected specimen. We have analyzed demographic, clinical and histopathological characteristics of the cases where diagnosis of DC was established or confirmed in the resected specimens. Our case collection comprising of 66 patients, is the largest case series on DCs

reported in the English literature till date. Previously reported series and their salient findings are summarized in Table 3.

Though DC is an anomaly which can present at any age, it is more commonly seen in children less than one year of age.<sup>3,6,9,10</sup> Of our patients, more than half (36/66, 54.6%) were infants. A slight male predominance has been noted by most authors and our findings corroborate with them.<sup>1,3,4,6,7,8,10,11,12</sup> In our cohort, the most common site was ileum, followed by jejunum and colon. Ileum is reportedly the most common location of DC, followed by esophagus and colon in other series.<sup>2,4,5,8</sup> In the colon, decreasing frequency is reported from caecum to rectum.<sup>2</sup>

Clinical presentation of DC is highly variable as it is related to certain factors like site of occurrence, size of lesion, type of mucosal lining and associated anomalies. Antenatal diagnosis by radiological imaging is not uncommon.<sup>3,4,9</sup> Two of our cases were picked during the course of antenatal routine ultrasound. Similarly, one case of Balakrishnan et al and ten out of 40 cases of Erginel et al were also diagnosed in utero.<sup>3,4</sup> An antenatal ultrasonogram can detect DC as early as 16th week of gestation.<sup>4</sup>

Presence of ectopic tissue such as gastric and pancreatic glands often produce misleading symptoms and can result in difficulties in clinical diagnosis. Gastric duplications frequently have ectopic pancreatic tissue and such patients may present with pancreatitis like symptoms with raised amylase levels (due to amylase secretion from the ectopic pancreatic glands) 5 as was the scenario in one of our cases. Common presenting symptoms include vomiting, rectal bleeding, abdominal lump, abdominal pain, feeding difficulty, constipation, cough, hemoptysis and respiratory distress.<sup>2,3,4,7,8,9</sup> Depending on the age at presentation and symptoms, the clinical differentials include perforation, intestinal obstruction, intussusception, appendicitis, peptic ulcer disease, pulmonary diseases, pancreatitis and even tumors.<sup>3,4,5,10</sup>

Patients with long standing DCs can present with complications like intestinal obstruction, volvulus, bleeding (due to ulceration or as a result of ectopic gastric mucosa), intussusception and perianal fistula (rectal DC).<sup>2,7</sup> An intussusception was noted in two of our cases and rectal bleeding in one. Anomalies which have been reported along with DCs include biliary obstruction, gastroschisis, vertebral anomalies and congenital cystic airway malformations.<sup>3</sup> Those noted in our cases include congenital diaphragmatic hernia, Meckel's diverticulum, ileal atresia, jejunal atresias and esophageal atresia.

With a clinical differential of myocardial infarction, one of our cases, a 47-year-old male with chest pain, was found to have an esophageal DC. Esophageal DC can be present in cervical, middle or distal esophagus in 23%, 17% and 60% cases of esophageal DCs respectively.<sup>4</sup> These can present as enlarging neck masses (upper esophagus),

respiratory symptoms (upper and middle esophagus) or remain asymptomatic (distal esophagus) with an often-incidental detection.<sup>4</sup>

Imaging studies play a significant role in diagnosis of DCs. Antenatal diagnosis, detection of incidental asymptomatic DCs and diagnosis of DCs not suspected clinically, are some of the scenarios which highlight the relevance of radiology in such cases. The different modalities which are useful include ultrasound, contrast enema examination, CT scan and MRI.<sup>2,5,6,8,9,11</sup> Often seen as an abdominal mass on ultrasonography or barium enhanced studies, DCs are characterized by an inner echogenic mucosa and outer hypoechogenic muscular layer. Contrast studies are useful for cases of communicating DCs and those associated with an intussusception.<sup>2</sup> If ultrasound findings are equivocal, CT scan and MRI can be employed. Smooth-walled rounded fluid-filled tubular or spherical cysts with mildly enhancing walls are seen on CT with a characteristic location adjacent to the gut. Tc-99 pertechnetate scan can pick up ectopic gastric mucosa and has been strongly recommended in children with occult or overt lower GIT bleeding.<sup>2,4,7</sup>

Based on their communication with the adjacent gut, DC can be communicating or non-communicating.<sup>4,5</sup> The spherical type of DC, which are the predominant category, do not communicate with the lumen and comprise nearly 82% of DCs. The other type, tubular DCs, constitute 18% of DCs, are more commonly seen in small and large intestinal locations and usually communicate with the lumen of the adjacent gut.<sup>4,5</sup> In our study also, there was a predominance of the non-communicating cysts (45/66, 68.2%).

Duplication cysts share the muscular wall and the blood supply with the adjacent gut whilst having a separate mucosal lining similar to or different from that of the adjacent gut.<sup>5</sup> The terminology is based on the part of gut to which DC is attached, rather than the mucosal lining of DC. Among ectopic mucosal linings, gastric is the most common (20-30%) and is commonly seen in esophageal DC, followed by small bowel DCs.<sup>3,5</sup> In our study group, gastric metaplasia was the commonest (27.3%), however, it was predominantly seen in ileal DC. Pancreatic tissue is another ectopic tissue found in DCs which is of clinical relevance. Macpherson et al reported pancreatic tissue in 37% of their gastric duplications.<sup>5</sup> Three of our cases, one each from duodenum, stomach and colon showed pancreatic heterotopia. Of our four esophageal DC cases, two showed respiratory lining.

Multiple theories have been proposed to explain the occurrence of DCs. The intrauterine vascular accident theory considers DC, similar to atresias, to be the result of a focal insult to the vascular supply of GIT owing to fetal stress and anoxia.<sup>2</sup> This theory holds its ground to explain those DCs which are associated with atresia.<sup>2,5</sup> Of our cases, 5 were associated with an atretic segment, three ileal and two jejunal. This theory fails to account for cases

where an associated atresia does not exist or where the atretic segment is located far away from the segment with DC, such as that seen in one of our cases where a one-month male presented with a rectal DC (with glial tissue) associated with an esophageal atresia. In a study by Macpherson et al, 9% patients were found to have associated atresias.<sup>5</sup>

Another common theory is the split notochord theory which postulates that development of a split notochord results in an aberrant connection between the yolk sac endoderm and the ectoderm which eventually produces a duplication.<sup>3-5</sup> It also successfully explains the vertebral anomalies associated with foregut DCs.<sup>5</sup> This theory can explain the occurrence of heterotopic glial tissue as seen in one of our cases with rectal DC. However, the associated esophageal atresia in the same patient is not fully explained by the split notochord theory alone.

The aberrant recanalization theory proposes that DCs result from incomplete recanalization of the gut as it passes from the solid stage to the luminal stage. This can be used to explain the communicating cysts seen in organs like esophagus, small bowel and colon, which have a solid stage of development, but not for other organs.<sup>4,5</sup> A plausible explanation for tubular colorectal DCs is abortive twinning theory, which also explains the association of genitourinary duplications with hindgut DCs, however fails to account for the communicating spherical duplication cysts.<sup>4,5</sup> The persistence of diverticula leading to produce DCs is the basis of persistent embryologic diverticula theory. Diverticula however, usually arise on the antimesenteric border as opposed to the commonly mesenteric location of DCs.<sup>4,5</sup> To date, no single hypothesis exists to satisfactorily explain the various locations, ectopic or metaplastic tissue and associated anomalies with duplication cysts.

This study is limited by the lack of follow-up of the patients. Hence a correlation between metaplasia and type or site of DC with patient outcome could not be made.

Once the diagnosis is established the primary treatment is complete surgical excision.<sup>4,8,9,11</sup> Additional mucosal stripping (Wrenn procedure) or partial removal of adjacent bowel segment may be required in cases when DC is communicating or is large in size.<sup>3,4</sup> Whole of the secreting or ectopic mucosa, including lining of DC must be removed so as to prevent recurrence and/or malignancies, in cases where complete removal of cyst is not feasible.<sup>4</sup> For patients who are not willing for a surgical procedure, a strict long-term follow-up should ensue owing to the potential risk of malignancies.<sup>4</sup>

## CONCLUSION

Duplication cysts are relatively uncommon entities which usually present in infants and young children. Their rarity and variability in clinical presentations makes clinical diagnosis elusive and often the diagnosis is clinched on

radiology and/or histopathology. Complete surgical excision is the mainstay of treatment. A high index of suspicion and knowledge of this congenital anomaly is helpful in suspecting the diagnosis, so that early diagnosis and appropriate management can ensue.

*Funding: No funding sources*

*Conflict of interest: None declared*

*Ethical approval: The study was approved by the Institutional Ethics Committee*

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**Cite this article as:** Aggarwal D, Vaiphei K. Gastrointestinal duplication cysts: experience from a tertiary care center in North India and review of literature. *Int Surg J* 2021;8:180-5.