



Gastrointestinal Duplications

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66.1 Introduction

Gastrointestinal duplications (GIDs) are rare, epithelium-lined congenital malformations that can arise anywhere in the gastrointestinal tract from the mouth to the anus. The mucosa frequently resembles the adjacent gastrointestinal tissue, but it can be heterotopic such as gastric or pancreatic tissue [1, 2]. It is the presence of this gastrointestinal mucosal lining that distinguishes GIDs from other gastrointestinal cysts [3]. Due to the rarity of these lesions and the variability of symptoms, GIDs frequently present both diagnostic and therapeutic challenges.

66.2 Demographics

Gastrointestinal duplications occur with a prevalence of 1:4500 births [4, 5]. Even though GID is rare, many reports exist with large numbers of patients accumulated over long periods [5–10]. There are some published reports of this condition from Africa [11–15].

About 60–70% of patients with GIDs present before the age of 2 years [5, 16]. There is a slight predominance in males [16]. In Africa, due to more difficulties with diagnosis, patients may present at an older age.

66.3 Embryology

Duplications are typically classified by their embryologic origin. The foregut consists of the pharynx, respiratory tract, and esophagus through the second portion of the duodenum. The midgut is the distal half of the second part of the duodenum through the proximal two thirds of the transverse colon, and the hindgut is the distal third of the transverse colon through the anus as well as the urologic system [3].

There are many theories regarding the embryologic origins of GID, but there is no single unifying theory that is able to explain all types of duplication. The presence of heterotopic tissue in duplications and the consistent mesenteric location of duplications have put many of these theories in question.

These embryological theories include the following:

- *Split notochord syndrome*: One of the most commonly proposed theories, it postulates that there is a space between the notochord and the endoderm during development through which the endoderm can herniate, create adhesions, and then persist to varying degrees. This may explain the dorsal location of duplications and the frequently associated spinal cord abnormalities [17].

- *Abnormalities of recanalization*: This theory postulates that the GI tract begins as a solid stage that later canalizes. Problems with this process may explain the formation of intestinal diverticula in embryos. However, this process occurs on both the mesenteric and antimesenteric sides of the bowel, whereas duplications are found only on the mesenteric side [3].
- *Remnants of embryologic diverticula*: Both intestinal diverticula in embryos and enteric duplications are coincidentally located most commonly in the ileum, indicating that perhaps enteric duplications are remnants of these embryologic diverticula. However, the presence of heterotopic mucosa, the mesenteric-only location of GIDs, and the presence of tubular duplications put this theory to question [3].
- *Partial twinning*: This theory posits that duplications are the result of partial twinning and is based on the association between GIDs and duplications of other body parts (head, trunk, extremities, gastrointestinal, and genitourinary tracts) [18].
- *Environmental factors*: This theory postulates that environmental factors such as trauma or hypoxia can lead to duplications, twinning, and intestinal atresia; duplications may be a part of the spectrum of intestinal atresia [18].

66.4 Pathology

In 1937, William E. Ladd coined the term *alimentary tract duplications*. He described three criteria for duplications: (1) an epithelial lining representing some type of intestinal tract mucosa, (2) a well-developed smooth muscle coat, and (3) intimate anatomic association with some portion of the gastrointestinal tract [19].

GIDs are most often spherical (■ Fig. 66.1), located in the ileum, singular, and contain mucosa that is similar to that of their adjacent gastrointestinal location (■ Fig. 66.2). [3] However, they may be tubular, be present anywhere in the gastrointestinal tract (■ Table 66.1), be multiple (as is the case in 10–20% of patients), and may contain heterotopic mucosa such as gastric or pancreatic [16]. They are always found on the dorsal or mesenteric side.

66.5 Diagnosis

66.5.1 Clinical Features

There is no common clinical pattern of signs and symptoms of duplications. The clinical presentation varies according to the age of the patient, location of duplication, type of mucosal lining, duration of disease, and presence of complications.



Fig. 66.1 Spherical duplication of the ileocecal area in an 8-month-old child

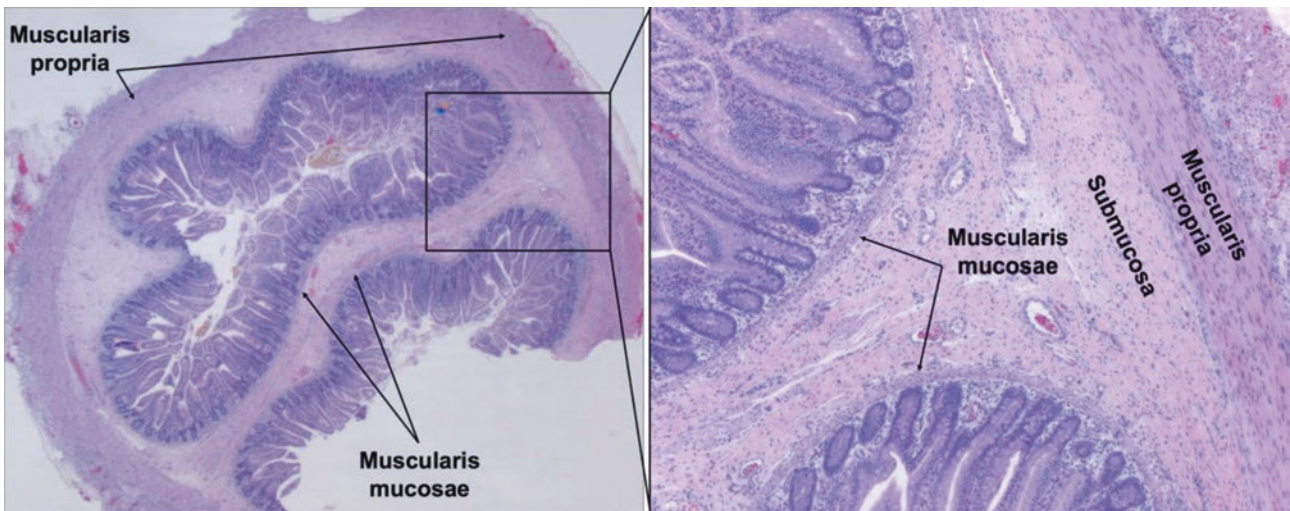


Fig. 66.2 Microscopic pathology of a gastrointestinal duplication demonstrating a duplicated mucosal layer with well-developed smooth muscle

Duplications on the floor of the mouth and foregut are associated with feeding difficulties. In thoracic duplications, patients have respiratory distress, wheezing, recurrent pneumonia, dysphagia, and failure to thrive. In the abdomen, GIDs most commonly cause vague abdominal pain. Mass effect from GIDs can lead to gastric outlet, intestinal, ureteric, biliary, or even vena caval obstruction as well as pancreatitis. Large presacral duplications can mimic sacrococcygeal tumors. GIDs can also present with intussusception, volvulus, perforation, bleeding (related to ectopic gastric mucosa), and malignant transformation [5, 16, 22, 23].

66.5.2 Prenatal Diagnosis

Prenatal diagnosis of GIDs is becoming more common in the Western world due to prevalence of prenatal ultrasound [24]. Ultrasound is especially successful for foregut duplications. Prenatal ultrasonography allows for early intervention in some cases. For example, in one patient with a large thoracic GID leading to mediastinal shift and hydrops, placement of a thoracoamniotic shunt allowed for resolution of the shift and hydrops [25].

Table 66.1 Summary of distribution of 503 duplications in 467 patients in eight studies

Location	Bower et al. [6]	Hocking et al. [7]	Holocomb et al. [8]	Stringer et al. [9]	Karnak et al. [10]	Pulgandia et al. [5]	Okur et al. [20]	Erginel et al. [21]	Totals (%)
Oropharyngeal	0	–	0	2	1	–	0	2	5 (<1)
Esophagus	15	8	21	15	7	–	1	0	67 (13)
Stomach	8	8	8	10	1	6	2	2	45 (9)
Duodenum	4	1	2	3	3	7	5	3	28 (6)
Jejunum/ileum	34	32	47	21	17	51	16	24	242 (48)
Colon	12	4	15	10	9	5	5	9	69 (14)
Rectum	2	5	5	6	2	4	1	0	25 (5)
Thoracoabdominal	1	2	3	6	2	0	2	0	16 (3)
Others	2	0	0	4	0	0	0	0	6 (1)
Total duplications/total patients	78/64	60/53	101/96	77/72	42/38	73/72	32/32	40/40	503 (100)

66.5.3 Postnatal Diagnosis

66.5.3.1 Physical Exam

Depending on the location of GIDs, patients may or may not have physical exam findings. Rectal and anal duplications can frequently be identified on exam [26].

66.5.3.2 Ultrasonography

US is the most common modality used for diagnosis of GIDs and should be the first choice. It classically shows an anechoic mass surrounded by a double-layered wall, consisting of an inner echogenic mucosa and outer sonolucent muscular layer (■ Fig. 66.3). When this double-layered pattern is present on US, a GID is confirmed and there is no need for further radiologic evaluation [27, 28].

66.5.3.3 Plain X-Rays

A plain chest X-ray (anteroposterior/lateral) is most helpful in detecting foregut duplications in the chest, which manifest as masses, asymmetry of vasculature, or unilateral hyperexpansion [29]. Plain abdominal X-ray may show evidence of intestinal obstruction but is often nondiagnostic.

66.5.3.4 Contrast Medium Studies

Contrast medium studies will not be able to image the GID itself unless it communicates with the gastrointestinal lumen (■ Fig. 66.4), but may reveal compression or displacement of the adjacent organ.

66.5.3.5 Computed Tomography (CT) or Magnetic Resonance Imaging (MRI)

A CT scan or MRI may be employed in difficult cases to assist with localization, especially in the thorax, pelvis, and retroperitoneum. A spinal MRI can be used to outline the relationship of the cyst with the spinal column and spinal canal.

66.5.3.6 Technetium 99^m Pertechnetate Scintigraphy Scan

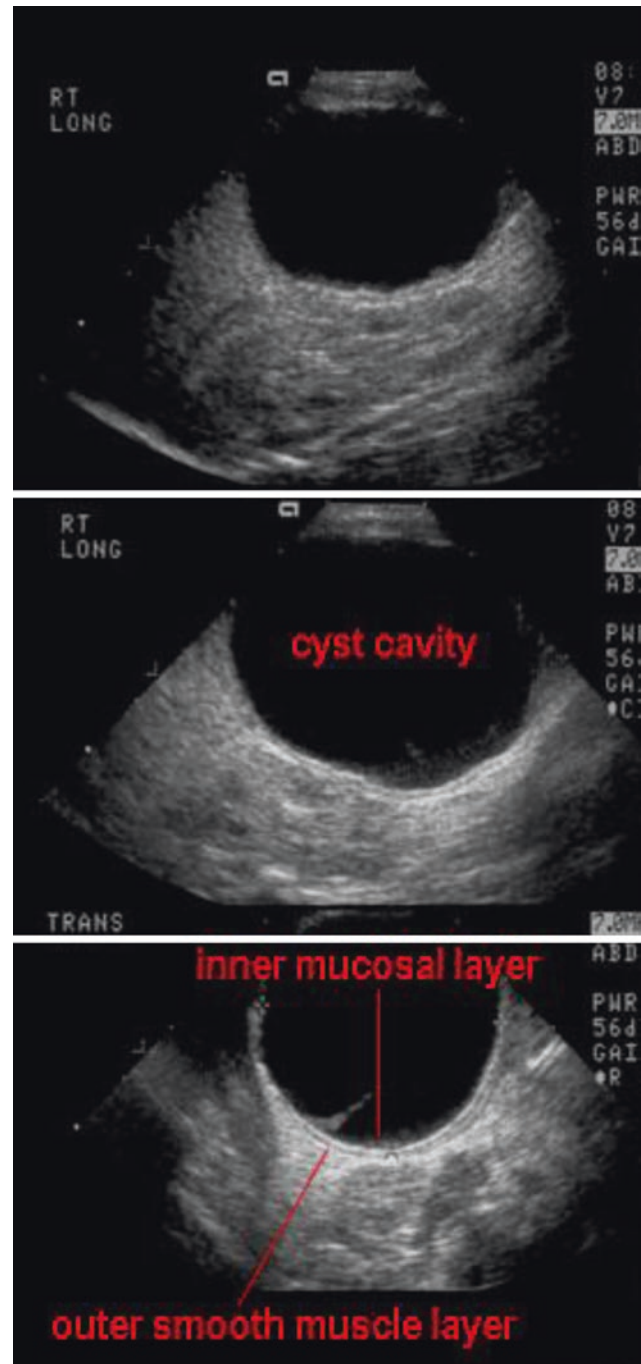
This scan indicates the definite existence of GID when it contains ectopic gastric mucosa. This is especially useful in esophageal, duodenal, and tubular small-bowel duplications with a high incidence of heterotopic gastric mucosa.

66.5.3.7 Laparoscopy

Laparoscopy is useful in cases when the above investigations are inconclusive.

66.5.3.8 Summary

In Africa, due to limited resources, CT scan and MRI are reserved for the very difficult cases. The most common investigations carried out are US and contrast medium examinations.



■ Fig. 66.3 Ultrasound images of a gastrointestinal duplication containing an inner mucosal layer and an outer smooth muscle layer

66.6 Treatment

The optimal approach to the care of children with GIDs is to make a prompt diagnosis and provide treatment before the onset of symptoms or the development of complications. When possible, GIDs should be completely excised. However, although malignant degeneration of GIDs is reported in adults, GID in itself is a



Fig. 66.4 Upper GI study demonstrating a tubular esophageal duplication

benign disease in children [8]. Therefore, any treatment should not be more radical than to eliminate the patient's symptoms and prevent further recurrence.

Important points to be considered in the surgical treatment of GID include the following:

1. The nature of the blood supply shared between the duplication and native bowel
2. The presence of heterotopic gastric mucosa, which will negate internal drainage due to the risk of peptic ulceration
3. The relationship with adjacent structures, such as the biliary tract in duodenal duplications

The treatment of GID is best considered by location of the duplication. However, in selected cases, an intraoperative frozen section may give further information on the absence or presence of heterotopic components.

66.6.1 Oropharynx

Oropharyngeal duplications are very rare lesions that may contain gastrointestinal ectopic mucosa. Though usually asymptomatic, these GIDs can cause significant respiratory and feeding difficulties and are usually diagnosed shortly after birth [30]. These cysts are excised by an intraoral incision.

66.6.2 Esophagus

Esophageal duplication cysts usually occur on the right side of the mediastinal esophagus [31]. Cervical esophageal duplications are rare, with less than 20 reported in the literature. Cervical GIDs are best approached through a cervical incision [32], whereas mediastinal GIDs require a right posterolateral thoracotomy or thoracoscopic approach if available [33].

66.6.3 Thoracic and Thoracoabdomen

These GIDs usually lie in the posterior mediastinum. They may cross the diaphragm and communicate with the proximal GI tract. They may also communicate with the spinal cord (called neuroenteric cysts). These connections can be missed in the OR, leading to high risk of complications [9]. In the case of neuroenteric cysts, resection should be performed in consultation with a neurosurgeon as incomplete excision can lead to meningitis. For isolated thoracic lesions, thoracoscopic resection is ideal, whereas thoracoabdominal cysts may require separate posterolateral thoracotomy and abdominal approaches in a staged fashion.

66.6.4 Stomach

Gastric duplications represent 2–8% of GIDs and are mostly located on the greater curvature [34]. They tend to be large and cause symptoms of mass effect, including gastric outlet obstruction and pancreatitis. They usually do not communicate with the stomach, and complete excision is possible. More extensive lesions are excised with a limited partial gastrectomy.

66.6.5 Duodenum

Duodenal duplications are rare, accounting for about 5% of GIDs. Total excision is not always possible if the cyst is in close proximity to the biliary or pancreatic ducts. The ducts may be included in the cyst walls, or the cyst and ducts may share the same blood supply. In these cases, the options include: [35]

- Partial resection of the cyst wall (as much as can be done safely) and resection of the entire mucosa
- Internal drainage through a wide cystoduodenal anastomosis or a cystojejunal Roux-en-Y anastomosis

66.6.6 Small Bowel

The small bowel (especially the ileum) is the most common site of all GIDs. Simple cystic GIDs can sometimes be shelled out, but most small intestine GIDs are resected with primary end-to-end anastomosis due to shared blood supply between the cyst and the bowel.

Tubular GIDs can sometimes involve the whole ileum and have an 80% incidence of gastric heterotopia; notably, unlike a Meckel's diverticulum, GIDs are located on the mesenteric side. Extensive resections can lead to short-bowel syndrome, and drainage into the adjacent normal bowel is not encouraged due to the risk of peptic ulceration. In these cases, the multiple stepwise stripping of the mucosa is as described by Wrenn [36], but anastomosis of the duplication to the stomach has also been attempted [37].

66.6.7 Colon

Colonic duplications rarely contain ectopic gastric mucosa. The rare complete colorectal duplication may be associated with doubling anomalies of the genitourinary organs, such as the bladder and vagina [38]. Cystic duplications can be shelled out or resected with primary colocolostomy. Tubular duplications of the colon have one or more communications with the native bowel. If the duplication is extensive and resection is not feasible, a distal communication is created with the colon or rectum to allow for drainage.

66.6.8 Rectum

Rectal duplications are usually presacral. In 80% of cases, there is no communication with the rectum, anus, or perianal skin. The differential diagnosis includes teratoma, neurofibroma, anterior meningocele, rectal leiomyosarcoma, and osteogenic sarcoma. Infection is common, and patients with small GIDs frequently undergo treatment for perirectal abscesses and anal fistulas with repeat recurrences [39]. The posterior sagittal approach is preferred for good exposure, safer removal and repair, and prevention of inadvertent compromise of the rectal lumen. For the rare anterior duplications, laparotomy can be performed.

66.6.9 Mesenteric Cysts

Mesenteric cysts can be confused with enteric duplications because they have similar presentations, and both are located on the mesenteric side of the alimentary tract. They can be found anywhere in the mesentery from the duodenum to the rectum, but they are most common in the small bowel mesentery [40, 41].

Like gastrointestinal duplications, the etiology of mesenteric cysts is unknown. They may arise from a disorder of the lymphatics such as misplaced lymph tissue, mechanical obstruction, or trauma. It has also been proposed that mesenteric cysts may be caused by failure of the mesenteric leaves to fuse or from formation of bowel diverticula which then grow in the mesentery as cysts [42].

Mesenteric cysts are thin-walled and contain either chyle (high triglyceride concentration) or serous fluid. Histologically, the cyst wall consists of fibrous tissue with an epithelial lining. Unlike gastrointestinal duplications, there is no mucosal or muscular layer [42].

Mesenteric cysts can be found incidentally by clinical palpation or ultrasound examination. They can also become symptomatic with abdominal pain, intestinal obstruction, or volvulus. Rupture can lead to the clinical picture of an "acute abdomen" with ascites and peritonitis.

The preferred treatment for these cysts is complete surgical excision that may require simultaneous segmental bowel resection [42].

66.6.10 Prognosis and Outcome

The outcome of surgical treatment of duplications is good. Poor outcomes are observed when there are associated severe congenital malformations, which in themselves carry a high morbidity and mortality.

66.7 Evidence-Based Research

GID is a very rare anomaly. As a result, while there are no large randomized trials on the diagnosis and management of this condition, many retrospective studies that are accumulated over time from referral centers are available. ■ Table 66.2 presents a large series of patients with GIDs, including their clinical presentations and location of each duplication within the gastrointestinal tract.

Table 66.2 Evidence-based research

Title	Gastrointestinal duplications
Authors	Puligandla PS, Nguyen LT, St-Vil D, Flageole H, Bensoussan AL, Nguyen V-H, Laberge J-M
Institution	The Montreal Children's Hospital, Montreal, Quebec, Canada; Hopital Ste Justine, Montreal, Quebec, Canada
Reference	J Pediatr Surg. 2003;38(5):740–744
Problem	This study characterizes the presentations of gastrointestinal duplications.
Study design	Retrospective study.
Length of study	22 years.
Results	Seventy-three patients with GIDs were treated during the study period. Most were identified as neonates and infants, but 24 presented after 1 year of age. Neonates and infants most often presented with vomiting and distension. Volvulus occurred in 23.8% of neonates. Children and adolescents tended to present with pain and vomiting. The most common GID location was the ileum (31.5%) and ileocecal valve (30.2%), followed by duodenum (9.6%), stomach (8.2%), jejunum (8.2%), colon (6.8%), and rectum (5.5%). Prenatal diagnosis by ultrasound has become more prevalent since 1991.
Outcome/effect	GIDs can present anywhere in the gastrointestinal tract, but they are most common in the ileum and can mimic Crohn's disease in older children. Prenatal ultrasound can help identify GIDs before the onset of symptoms.
Conclusions	GIDs have variable presentations and can lead to life-threatening complications. Early identification and treatment are critical.

Key Summary Points

1. GIDs are rare and can arise anywhere from the mouth to the anus.
2. The embryology of GIDs is unknown.
3. About half of all GIDs are located in the small bowel, with the ileum being most common.
4. The clinical features of gastrointestinal duplications are nonspecific; a high index of suspicion is required for prompt diagnosis.
5. Most patients with gastrointestinal duplications will present before 2 years of age, but presentation during adulthood, as well as late malignant transformation, has been described.

6. Ultrasonography should be the first line of investigation.
7. The ideal treatment is total excision.
8. The results of surgical treatment are good.

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