Journal of Clinical and Experimental Gastroenterology

Losefsky, J Clin Exp Gastroenterol. 2022;1(1):1-4.

Mini Review

A comprehensive review of enteric duplication cysts, their pathophysiology, presentation, and treatment

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Received date: April 05, 2022
Accepted date: May 03, 2022

Citation: Losefsky Q. A comprehensive review of enteric duplication cysts, their pathophysiology, presentation, and treatment. J Clin Exp Gastroenterol. 2022;1(1):1-4.

Abstract

Enteric duplication cysts are rare congenital malformations of the gastrointestinal tract that can be found anywhere along the entire alimentary canal. Duplication cysts share a muscular layer with the adjacent bowel and contain their own mucosal lining which may be that of any part of the alimentary tract. They are commonly discovered in the neonatal period and are rarely diagnosed after the second decade of life. This article discusses the etiology, presentation, diagnosis, and management of duplication cysts along all areas of the gastrointestinal tract.

Keywords: Enteric duplication cyst, Surgical oncology, Congenital malformation, Alimentary duplication cyst, Embryology

Introduction

Alimentary, or enteric, duplication cysts (EDC) are rare congenital anomalies, found anywhere along with alimentary tract from the tongue to the anus [1]. The term alimentary tract duplication was first described by Ladd in 1937 in an attempt to describe both the pathological and clinical nature of these lesions, as well as to merge a multitude of separate entities that had been described previously in the literature [2]. Most of these cysts are found in infants or children, although they can be found in any age group.

Gastrointestinal duplications are believed to occur with an incidence of 1 per 4000-500 live births, and most are found in infants or children. They are most commonly found in the jejunum (52%), esophagus (17%), colon (14%), stomach (8%), duodenum (6%), and rectum (6%) [3]. They very rarely can be found in remote areas such as the tongue, pleural space, retroperitoneum, pancreas, liver, and biliary tree [1]. Currently, there is no evidence that shows an association of duplication cysts with sex, race, or genetic predisposition [2].

EDCs are defined based on specific gross and histologic criteria. There are two types of duplications: tubular where the lumen is contiguous with the alimentary tract lumen, and cystic where there is no communication between lumens [4]. This review will focus on cystic duplications only. Grossly they are spheric cysts that are located in or immediately adjacent to part of the gastrointestinal tract and tend to be adhered to the mesenteric aspect. Additionally, they often share both a common muscular wall and blood supply, but have separate mucosal lining [1]. Histologically, they contain smooth muscle in their walls and have a lumen lined with alimentary tract mucosa. The mucosa does not necessarily have to be that of the adjacent segment of gastrointestinal tract, rather there can be a multitude of ectopic tissues such as gastric, pancreatic, ganglion cells, and lymphoid aggregates that resemble Peyer patches [1]. As such, duplication cysts are named for their location rather than their tissue type.

This review will discuss the current theories for etiology, details about presentation and unique attributes of duplication cysts found along all parts of the alimentary tract, diagnostics, differential diagnoses, and current management guidelines.

Etiology

There are multiple theories for the etiology of alimentary tract duplications. Briefly, the gut tube is formed from endodermal lining of the yolk sac which then becomes enveloped by mesodermal lining. The endoderm develops into the mucosal and submucosal structures while the mesoderm gives rise to the vascular supply, connective tissues, and muscular layers [2]. Finally, the enteric nervous system arises from the systematic migration of neural crest cells into the submucosa.

The split notochord theory hypothesizes that there is an incomplete separation of the notochord from the gastrointestinal endoderm, and is commonly used to explain thoracic duplications [3]. Esophageal duplications in particular have a high prevalence of associated vertebral anomalies, which makes this theory favorable. If this were true, then it would infer that alimentary tract duplications are neurenteric cysts, which is not generalizable to the whole of duplications [1].

Another hypothesis involves recanalization errors involving the neonatal solid GI tract during weeks 7-8 of embryogenesis [3]. This theory suggests that there is some disruption in the process of recanalization that then allows a duplication to occur. Unfortunately, this theory would only account for those areas of the GI tract which undergo recanalization.

The "abortive twinning" theory proposes that enteric duplications could be a result of incomplete twinning. This theory could explain the subsect of colorectal tubular duplications or "duplications of the hindgut" that are associated with duplications of genital and urinary structures [1]. While it could account for this type of duplication, this theory does not account for most gastrointestinal tract duplications, particularly the spherical cysts at any level.

An alternative hypothesis is the "persistent embryologic diverticula" theory which suggests that small, usually temporary, diverticula found on the antimesenteric aspect of embryo intestinal walls persist and develop into enteric duplications [1]. Most enteric duplications develop on the mesenteric side, which would not be accounted for with this theory. Additionally, this etiology would only be plausible for intestinal duplications.

Finally, the intrauterine vascular accident theory suggests that duplications arise due to intrauterine vascular accidents, much like the etiology of small bowel atresias. There have been a number of cases of enteric duplication cysts associated with intestinal atresias, most often in the same segment of the gastrointestinal tract [1]. This association demonstrates some level of association between duplications and atresias at all levels of the alimentary tract, but the theory is only a valid explanation for duplication cysts associated with an atresia.

General Imaging and Diagnosis

Ultrasonography is one of the most used modalities to assist in the identification of enteric duplications [2]. On ultrasound, cystic duplications should show an anechoic fluid-filled double-walled cyst composed of an inner hyperechoic rim of mucosa-submucosa and an outer hypoechoic layer of smooth muscle which is consistent with the muscularis propria [3]. This double layered appearance of the mass is often referred to as the "gut wall signature" and usually is not seen with other intra-abdominal cysts or abscesses [2].

Computed tomography (CT) imaging with oral contrast may be helpful as the contrast will not fill the cystic duplication, but may demonstrate a compression effect on adjacent structures [3]. Additionally, CT or magnetic resonance imaging (MRI) can provide some additional information and demonstrate the relationship between the cyst and the adjacent bowel. In the absence of significant findings on imaging, endoscopy or laparoscopy can be a useful diagnostic adjunct [2].

Esophageal Duplication Cysts

EDCs of the esophagus comprise about 10-15% of all duplication cysts; 60% are located in the lower third of the esophagus, 17% in the middle third, and 23% in the upper third [5]. The prevalence of these cysts is estimated to be 0.0122% and as many as 80% of these lesions are diagnosed in childhood, the majority being symptomatic [6]. About 43% of esophageal EDCs contain ectopic gastric mucosa, which can lead to peptic ulcers [1]. Additionally, vertebral anomalies and esophageal atresias seem to be the most prevalent congenital anomalies associated with esophageal duplication [1]. EDCs in the esophagus may develop symptoms such as dysphagia or chest pain due to mass effect, and can also cause stridor or a nonproductive cough depending on the location of the cyst [6].

Duplications of the cervical portion of the esophagus usually present as an asymptomatic, enlarging lateral neck mass or with symptoms of upper airway obstruction in the newborn period [1]. Differential diagnoses with this presentation include a thyroglossal duct cyst, branchial cleft cyst, cervical lymphadenopathy, cystic hygroma, and a variety of solid cervical tumors.

EDCs in the middle third of the esophagus are in close proximity to the intrathoracic trachea, so often generate symptoms of severe upper airway obstruction in early infancy. The differential diagnoses include bronchogenic cysts, neurenteric cysts, malignant mediastinal masses, and various intramural tumors of the esophagus [1]. If there is concern for malignant causes of mediastinal masses, additional lab tests could be ordered. α-FP and β-hcg for seminoma and non-seminomatous germ cell tumors, lactate dehydrogenase (LDH) for lymphoblastic non-Hodgkin lymphoma, serum alkaline phosphatase (ALP), and white blood cell count for Hodgkin disease/mediastinal large cell lymphoma [7]. In all cases, a surgical specimen with histopathology is required for a definitive diagnosis.

Finally, the remaining 60% of esophageal EDCs can be found in the distal third. In this location, EDCs are frequently asymptomatic and are often found incidentally on routine imaging [1]. The differential diagnosis for this location is the same as that in the middle third of the esophagus.

In the past, esophageal duplications have been managed with marsupialization followed by the destruction of the mucosa with curettage or gauze packing [8]. This method was found to cause many complications and need for repeat surgical management and has since been mostly abandoned in favor of complete excision. There have been cases of large, thoracoabdominal tubular cysts that were firmly adhered to the esophagus and so management was deferred to the marsupialization method [8].

Gastric Duplication Cysts

Duplication cysts of the stomach make up between 4-9% of all intestinal duplications [6]. Like duplication cysts found elsewhere,

the majority are found in childhood or infancy. Gastric duplications often present with symptoms of obstruction, abdominal pain, or a palpable mass [1]. More than 80% of gastric duplications are cystic in nature and do not communicate with the lumen [4].

Most EDCs of the stomach are located along the greater curvature, only 5.5% are found on along the lesser curvature [6]. These cysts may be adherent to the pancreas and could even communicate with aberrant pancreatic ducts. Ectopic pancreatic tissue is most commonly found in gastric duplications (37%), and may be associated with pancreatitis and elevated amylase levels [1]. Patients may also develop symptoms such as diffuse abdominal pain, epigastric pain, vomiting, weight loss, or gastric outlet obstruction. The differential diagnosis for this presentation includes pancreatic cyst and pseudocyst, mesenteric cyst, and various intramural tumors of the stomach.

The management of these cysts is generally recommended to be complete excision of the duplication without violation of the gastric lumen. If this is not possible, segmental gastrectomy may be necessary [8].

Biliary Duplication Cysts

EDCs of the biliary tract are exceedingly rare, and only account for less than 1% of all duplication cysts. Most are identified during childhood, and present with a vague history of postprandial epigastric pain, and a number of patients have transient episodes of recurrent jaundice [9].

Diagnosis in adults is challenging, both because of the rarity of this condition as well as a broad differential diagnosis. Biliary duplication cysts have commonly been mistaken for choledochal cysts, double gallbladder, sarcoma, or even cholangiocarcinoma. [9]. A Tc-99 pertechnetate scintigraphy can be used to identify the presence of ectopic gastric mucosa in cases suspicious for a duplication cyst, and cholangiography can locate the site of obstruction and communication with the biliary tree [9].

Due to the difficult location of these cysts, complete open resection is the current treatment of choice rather than a laparoscopic approach [9].

Small Bowel Duplication Cysts

Alimentary tract duplications of the small bowel can be found in all three sub-sections of the small bowel. Jejunal duplications comprise of about 50% of gastrointestinal tract duplications, ileal about 44%, and duodenal anywhere from 2-12% [6]. Like other duplications, those in the small bowel generally contain two mucosal layers and share a common muscular wall. One third of all small bowel duplications are symptomatic in the neonatal period, and 72% have a clinical onset before the patient is 2 years old [1].

Depending on the location of the EDC, patients can present with a variety of symptoms including vomiting, abdominal pain, even melena. Duodenal cysts can cause other complications such as pancreatitis, weight loss, and gastrointestinal bleeding from ulceration of ectopic gastric mucosa within the cyst [6]. Jejunal duplication cysts can present with abdominal bloating, constipation, intussusception, volvulus, and partial small bowel obstruction. Finally, ileal duplications can present with small bowel obstruction, palpable abdominal mass, or hematochezia.

A subtype of small bowel duplications that is not recognized by all authors is the ileocecal duplication. These are in the most terminal portion of the ileum or on the ileocecal valve itself. The cyst is usually located on the mesenteric border of the bowel and have an association with small bowel atresia. Ectopic gastric mucosa can be found in up to 24% of these cysts [1].

The differential diagnosis for small bowel EDCs includes all causes of neonatal bowel obstruction and intussusception. Additionally, mesenteric or omental cysts, pancreatic pseudocysts, exophytic hepatic cysts, and in females, ovarian cysts should be considered.

Much like other EDCs, a complete resection of the duplication is the preferred management. If a complete excision is not possible, it is important to remove the mucosa of the duplication due to the potential of retention of heterotopic tissue. Retained ectopic gastric mucosa could lead to peptic ulceration for example [8]. Advances in endoscopy has seen some success with endoscopic marsupialization, but there is still concern for malignancy or bleeding form peptic ulceration [3]. While the treatment of asymptomatic cysts is still somewhat controversial, it is generally recommended to do a complete surgical resection as there is the potential for future complications.

Colonic/Rectal Duplication Cysts

Duplication cysts of the colon and rectum represent 6.8% of alimentary tract duplications. These duplications can be located anywhere in the large intestine, and may present as abdominal pain, acute abdomen, obstruction, and/or gastrointestinal bleeding [6]. They are divided into three subtypes: duplication of the appendix, cystic duplications of the colon, and colorectal tubular duplications. Duplications of the appendix or "double appendix" are usually incidental surgical findings and will not be discussed further. Additionally, colonic tubular duplications seem to be a distinctly different entity with a separate pathogenesis when compared to duplications in other parts of the alimentary tract and therefore will not be included in discussion [1].

Cystic colonic duplications generally account for around 50% of large intestinal duplications, and are often found on the cecum [1]. Cystic duplications of the rectum account for about 4% of gastrointestinal duplications, usually located posterior to the rectum or anus. While they can occur at any age, most large intestine duplications are discovered during infancy. Common presenting symptoms include fecal soiling, constipation, and a palpable retrorectal or retroanal mass on digital examination [1]. The differential diagnosis includes anterior meningocele, sacrococcygeal teratoma, retrorectal abscess, pilonidal cyst, and sacral bone tumors.

While malignant degeneration of duplication cysts can occur at any level of the enteric tract, it is most often reported in the colon. Up to 67% of malignancies diagnosed in duplication cysts occur in the colon [6]. Management of symptomatic cysts is generally complete surgical excision. Routine surgical resection of these cysts is recommended if patients are asymptomatic as well due to the potential for perforation, bleeding, obstruction, and malignant degeneration [6]. Surgical resection usually involves a partial colectomy with complete cyst resection in good surgical candidates.

Conclusions

Alimentary tract duplications are a relatively rare finding, occurring in 1 in every 4000-5000 births [3]. They can be found

at any level of the alimentary tract and are usually discovered before the age of 10. Histologically, duplication cysts share a muscular layer with the adjacent bowel and have a separate mucosa which usually resembles nearby bowel mucosa but may also contain heterotopic alimentary mucosa.

While there have been multiple theories regarding the etiology of EDCs, no single theory has proven satisfactory. Duplications at all levels of the gastrointestinal tract appear to share enough characteristics that a single etiologic explanation should be possible, but none proposed thus far appear to explain all duplications in all parts of the alimentary tract.

In general, surgical removal or enucleation is the treatment of choice for most, if not all, EDCs [6]. There is some debate regarding the preferred management in asymptomatic duplications. On one hand, surgical removal of asymptomatic cysts could be considered as there is a risk of future complications such as malignant transformation, ulceration, or perforation, and short-term postoperative outcomes in these patients has so far been excellent [6]. On the other hand, surgery always carries a certain amount of risk and there has been some success with observation with serial ultrasounds.

Between 1955 and 2012, there were 64 citations in the literature that provide adequate descriptions of 67 cases of malignancies arising from alimentary tract duplications near the esophagus, stomach, small intestine, appendix, and large intestine [10]. 57 of these cases underwent surgical treatment, and short-term outcomes were favorable [10]. In this series, 43 patients had known prognosis, 7 died from tumor progression and in another 5 cases the tumors had recurred and metastasized after surgery at an average of 11.4 months. This case series suggests that all duplications should be considered for surgery due to the possibility of malignant transformation, and the poor prognosis once malignancy has occurred in these duplications [10].

Further study involving the risks and benefits of surgical resection of asymptomatic EDCs is required to ascertain best practice. Unfortunately, due to the rare nature of this congenital malformation, it is difficult to complete such studies.

Abbreviations

EDC: Enteric Duplication Cyst; CT: Computed Tomography; GI: Gastrointestinal; MRI: Magnetic Resonance Imaging

All Financial Interests (Including Pharmaceutical and Device Product(s)

There was no funding organization or sponsor: “design and conduct of the study: none; collection, management, analysis, and interpretation of the data: none; preparation, review, or approval of the manuscript: none, and decision to submit the manuscript for publication: none”

Ethical Approval

Exempt from IRB review.

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