Duplication Cyst

Sachit Anand

Abdul Aleem MD
Lehigh Valley Health Network, Abdul.Aleem@lvhn.org

Published In/Presented At

Anand S, Aleem A. Duplication Cyst. 2022 May 4. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan–. PMID: 33232017.

Duplication Cyst

Anand S, Aleem A.

Continuing Education Activity

Duplication cysts are rare congenital anomalies of the alimentary tract. The majority of duplication cysts are symptomatic within the first 2 years of life. Due to the varied clinical presentations, radiological evaluation is required for its diagnosis. This activity reviews the evaluation and treatment of duplication cyst and highlights the role of the interprofessional team in the care of patients with this condition.

Objectives:

- Explain the etiology of duplication cysts.
- Describe the clinical presentation and typical imaging findings associated with duplication cysts.
- Outline the common approaches to surgical management of children with duplication cysts.
- Summarize the importance of interprofessional team strategies for improving care coordination and communication to aid in the precise diagnosis of duplication cyst and improve outcomes in patients diagnosed with this condition.

Access free multiple choice questions on this topic.

Introduction

Duplication cysts, also known as alimentary tract duplications, are congenital lesions of the gastrointestinal tract. Although the exact cause still eludes us, multiple theories have been put forward to explain its occurrence.[1][2] This rare anomaly has an incidence of 1 in 4500 births with a slight male preponderance.[3] The majority of these cysts are symptomatic within the first 2 years of life. Even with the advances in prenatal imaging, a prenatal diagnosis on ultrasound is possible in only 20% to 30% of the cases.[1]

A diverse clinical presentation due to variations in location, size, presence of heterotopic mucosa, etc. makes the clinical diagnosis of these cysts challenging. Ultrasound showing the gut signature sign is pathognomonic of a duplication cyst. The definitive treatment modality for children with a duplication cyst is surgery. Excellent long-term outcomes are reported after optimal surgery.

Etiology

Although the exact cause of alimentary tract duplications is not known, these are believed to occur due to embryological aberrations between 4th to 8th weeks of gestation. Multiple theories have been put forward to explain its etiopathogenesis. The split notochord theory is based on the abnormal separation of the growing notochord from the endodermal cells. This theory is mostly cited to explain the occurrence of vertebral anomalies associated with duplication cysts.[1]

The recanalization theory postulates that errors in the recanalization of the gut are responsible for the occurrence of these cysts.[1] However, it fails to explain the occurrence of duplications in the areas which do not undergo the process of recanalization during their embryological development. The embryonic diverticula theory is based on the explanation that there exist some diverticulae in the growing embryo. A persistent diverticulum will grow with the growth of the alimentary tract and will contribute to the genesis of alimentary tract duplication. Although it gives a fair justification of the mechanism behind the occurrence of the majority of the duplication cysts, it fails to explain the reason for heterotopic mucosa in these cysts.[1][2]

The partial or abortive twinning theory explains the occurrence of these duplication cysts is due to incomplete twinning of the alimentary tract.[1] The association of doubling anomalies of the genitourinary tract with colorectal duplication cysts clearly justifies the theory; however, it fails to explain the occurrence of these cysts in other areas.

Epidemiology

The incidence of alimentary tract duplications is 1 in 4500 births. A slight male preponderance has been observed with these cysts.[3] They are mainly detected in childhood, with the majority of duplications symptomatic within the first 2 years of life. However, late presentation in adulthood is also seen.[4]

Pathophysiology

Three distinct characteristics seen in these cysts include an epithelial lining representing a part of the gut, a well-defined layer of smooth muscle, and a close approximation with some part of the gastrointestinal tract, such as sharing a common wall.[1]

Cysts can be divided into cystic or tubular on the basis of their structural configuration. Cystic duplications are more common (80% of the duplications) and do not communicate with the bowel lumen. Tubular duplications are less common and communicate with the lumen of the bowel. [4]

These cysts can be divided into foregut, midgut, and hindgut duplications depending on the part of the alimentary tract they are intimately attached to. The ileum, followed by the esophagus, is the most common location for these duplication cysts.[1] The foregut duplications can be further divided into esophageal, bronchogenic, and neurenteric, depending on their embryological origin.[4]

Histopathology

On histopathology, a distinct mucosal lining and smooth muscle coat are characteristic features of these cysts. The mucosal lining generally corresponds to some part of the gastrointestinal tract. The mucosal lining may be heterotopic and may not correlate with the adjacent bowel. Ectopic gastric mucosa is seen in approximately 20% to 30% of the cases and is common in esophageal and midgut duplication cysts. Pancreatic mucosa is commonly observed in gastric duplications. Besides this, bronchogenic cysts have respiratory epithelium, cartilages, and bronchial submucosal glands. [1][4]

History and Physical

The majority of the cases having an alimentary tract duplication present within childhood. The type of symptoms depends on the location of the cyst, type of the cyst (cystic or tubular), and presence or absence of ectopic mucosa. Patients with cystic duplications are generally symptomatic with abdominal pain but can also be incidentally detected. Partially communicating (with only one end communicating with the adjacent bowel), tubular duplications are known to cause symptoms such as chronic constipation due to the loading of the intestinal contents in a blind loop. Cysts with heterotopic gastric or pancreatic mucosa present as abdominal pain and gastrointestinal bleeding due to ulceration and bleeding of the healthy adjacent bowel.

Clinical presentation based on the location of the cyst

Foregut duplication cyst: Dysphagia, vomiting, epigastric pain, and upper gastrointestinal bleeding are common clinical manifestations of esophageal duplication cysts. They can also cause compression on the surrounding structures and present as a large neck mass, stridor, dyspnea, cough, etc. Patients with bronchogenic cysts also present with similar features of dysphagia, chest pain, cough, dyspnea, etc. Foregut duplication cysts can also be prenatally detected on the routine ultrasound. Features, including intrathoracic mass, polyhydramnios, mediastinal shift, and hydrops, can be observed. Gastric duplication cysts are rare and usually present with abdominal pain, vomiting, abdominal mass, and features of gastric outlet obstruction. [4] Duodenal duplication cysts may present as abdominal pain, nausea, vomiting, jaundice, and upper gastrointestinal bleeding. Rarely, they may present with features of acute pancreatitis.[5]

Midgut duplication cyst: Children with small bowel duplications usually present with abdominal pain, palpable mass, nausea, and vomiting. They may present with features of upper gastrointestinal bleeding due to heterotopic mucosa. Acute abdominal pain due to intussusception or volvulus is also reported in the literature.

Hindgut duplication cyst: Colonic duplication cysts are rare as compared to small bowel duplication cysts. They usually contain colonic mucosa; however, some cases with ectopic mucosa have also been reported. Similar to small bowel duplication cysts, these may present as abdominal pain, palpable mass, vomiting, gastrointestinal bleeding, or features of intestinal obstruction. Malignant transformation has also been seen in these cysts. In addition, rectal duplications can present with distinct clinical presentations, including presacral masses, imperforate anus with rectovaginal fistula, etc. [4][6]

Evaluation

A prenatal diagnosis from an ultrasound examination is possible for these lesions. However, the sensitivity is only 20% to 30%.[1] Due to a varied clinical presentation, radiological investigations play a vital role in the diagnosis of duplication cysts. Ultrasound of the abdomen shows a classical five-layered cyst wall with alternating hyperechoic and hypoechoic layers. This is known as the gut signature sign and is pathognomonic for alimentary tract duplication. Although ultrasound can also be used to diagnose foregut duplications located in the neck, its sensitivity is poor for intrathoracic lesions. Transesophageal ultrasound (TEE) can be used for these lesions, but it is not readily available at all centers, especially in developing countries. Contrast-enhanced computed tomography (CECT) can be used to localize the lesions in these scenarios. On CECT, these

duplications appear as hypoattenuating masses with an enhancing rim. Similar to intrathoracic lesions, it might be difficult to localize and diagnose rectal duplication cysts located deep in the pelvic cavity. Magnetic resonance imaging (MRI) provides better cross-sectional anatomy in these cases and also helps in the diagnosis of duplication cysts involving the hindgut.[1]

A technetium-99m pertechnetate scan can be done to know whether the cyst contains ectopic gastric mucosa or not. Although it might not change our management, it can help in diagnosing multiple synchronous lesions. It is better to have knowledge about the duplication cysts that will rarely show a positive pertechnetate scan like bronchogenic and rectal duplication cysts. This radionuclide scan can, therefore, be avoided in these cases.

Treatment / Management

The definitive treatment for a duplication cyst is surgery. The surgical approach can be open (laparotomy or thoracotomy) or minimally-invasive (laparoscopic or thoracoscopic) and depends on the expertise of the surgeon.

Management of antenatally or incidentally diagnosed asymptomatic lesions: Antenatal ultrasound can diagnose these anomalies, and early surgery should be done. This is not only due to malignant transformation, but also, if left untreated, a significant proportion of these cases may develop complications such as intussusception, volvulus, hemorrhage, etc.[1]

Symptomatic cases: Early surgery is recommended in all the symptomatic cases.

Intrathoracic esophageal duplication cysts: These require a posterolateral thoracotomy. These can be removed by simple excision. In large cysts, decompression of the cyst may help in the better dissection of the cyst.

Gastric and intestinal duplication cysts: Laparotomy with excision of the cyst can be done in the majority of cases. Resection with end-to-end anastomosis might be required in tubular duplications or those intimately associated with the bowel wall. Long tubular duplications or multiple duplications involving the small intestine can be removed by performing mucosal stripping or Wrenn's procedure.[7] Similarly, long tubular duplications of the colon can be taken care of by performing Soper's procedure.[8] Communication is established between the distal blind duplication and healthy bowel for the evacuation of stools.

Rectal duplication cysts: These cysts might present as presacral masses or imperforate anus with rectovaginal fistula. Therefore, these may require a posterior sagittal or abdominoperineal approach for cyst excision.

Special scenarios:

Synchronous cysts in the thorax and abdomen: Synchronous lesions are seen in 10%-15% of the cases.[9] In the condition of synchronous cysts, it is valid to excise the symptomatic one first. However, if both are symptomatic, then the thoracic cyst can be excised first to minimize the anesthesia-related issues. It must be kept in mind that with modern anesthesia, both of the cysts can be excised in a single sitting. However, this increases morbidity leading to the risk of postoperative mechanical ventilation, the prolonged requirement of analgesics, and prolonged hospital stay, etc.

Neurenteric cysts: A proper evaluation with MRI of the spine and surgical planning with the neurosurgery team is crucial. If the intraspinal component is significant, the intraspinal portion should be removed first.

Differential Diagnosis

The common differential diagnosis for alimentary tract duplications include:

- Meckel's diverticulum: It presents in a similar age group and has a similar presentation of abdominal pain and upper gastrointestinal bleeding. A pertechnetate scan can be positive in both scenarios. However, a definite diagnosis is established only on exploration. The distinct difference is location. Duplication cysts are located on the mesenteric side, while Meckel's diverticulum is located on the anti-mesenteric side.
- Congenital segmental intestinal dilatation: It is characterized by a segmental dilatation of the small or large intestine up to three to four times. Although dilatation can be seen in both duplication cysts and congenital segmental dilatation, the difference between these two is the lack of any cystic structure adjacent to the normal bowel in segmental dilatation. However, the treatment of both is similar.[10]
- Other intraabdominal cystic lesions: These include mesenteric, omental, choledochal, and ovarian, etc.
- Presacral masses: Rectal duplication cysts can mimic other presacral masses, including sacrococcygeal teratoma, anterior meningocele, and dermoid, etc. Magnetic resonance imaging and biochemical tests, including alpha-fetoprotein assay, usually help in definite diagnosis.
- Other intrathoracic cysts: Other intrathoracic cysts, including the pericardial cyst and the thymic cyst, may be considered as close differentials of duplication cysts. However, these are located in the middle and the anterior mediastinum, respectively, unlike the duplication

cyst, which is located in the posterior mediastinum.

Prognosis

Alimentary tract duplication cysts are commonly seen in children. Surgical resection is required in the majority of cases. However, a proportion of cases may require complex surgeries. Excellent long-term outcomes are reported after optimal surgery.

Complications

Delayed diagnosis and surgery can lead to complications, including volvulus, intussusception, recurrent hemorrhage, and risk of malignant degeneration, etc.[1] Perforation of large duplication cysts has also been reported in the literature.[11] Complications after surgery are similar to any other laparotomy, including a spectrum ranging from localized wound infection to anastomotic leak and adhesive obstruction.

Consultations

Children with alimentary tract duplications require the involvement of multiple disciplines for its optimal management. A radiologist is involved in the multidisciplinary team right from the antenatal diagnosis of a duplication cyst. The involvement of a gynecologist and fetal surgeon is necessary in case fetal intervention is required due to severe polyhydramnios and hydrops. The radiologist also needs to be consulted for cysts presenting in the postnatal period. A primary clinician or neonatologist plays a crucial role in the preoperative management of the child. A pediatric surgeon needs to be consulted for the surgical management of the child. The involvement of a neurosurgeon is also crucial in cases requiring the management of cysts with intraspinal extension.

Deterrence and Patient Education

Parents must be educated about the various approaches to surgical management. In cases that are antenatally diagnosed, the couple must be informed about the symptoms that can occur after birth, various modalities of diagnosis, and approaches to management. Information about the possible surgical complications must also be provided to the parents.

Enhancing Healthcare Team Outcomes

Management of duplication cysts requires an interprofessional team approach. The involvement of a pediatric surgeon, primary clinician or neonatologist, radiologist, gynecologist, and neurosurgeon are necessary in the optimal management of these cases. The nurses are also a vital member of the interprofessional group. They play an important role in the postoperative monitoring and care of the children. The pharmacist must ensure that the patient is on the correct formulation and doses of antibiotics and analgesics in the postoperative period. Thus, meticulous planning and discussions with all the team members involved in the management of the patient are highly recommended to lower morbidity and improve outcomes. [Level 5]

Review Questions

- Access free multiple choice questions on this topic.
- Comment on this article.

References

1. Sangüesa Nebot C, Llorens Salvador R, Carazo Palacios E, Picó Aliaga S, Ibañez Pradas V. Enteric duplication cysts in children: varied presentations, varied imaging findings. Insights Imaging. 2018 Dec;9(6):1097-1106. [PMC free article: PMC6269332] [PubMed: 30311079]
2. Pant N, Grover JK, Madan NK, Chadha R, Agarwal K, Choudhury SR. Completely isolated enteric duplication cyst associated with a classic enterogenous duplication cyst. J Indian Assoc Pediatr Surg. 2012 Apr;17(2):68-70. [PMC free article: PMC3326825] [PubMed: 22529551]
3. Tiwari C, Shah H, Waghmare M, Makhija D, Khedkar K. Cysts of Gastrointestinal Origin in Children: Varied Presentation. Pediatr Gastroenterol Hepatol Nutr. 2017 Jun;20(2):94-99. [PMC free article: PMC5517385] [PubMed: 28730133]
4. Liu R, Adler DG. Duplication cysts: Diagnosis, management, and the role of endoscopic ultrasound. Endosc Ultrasound. 2014 Jul;3(3):152-60. [PMC free article: PMC4145475] [PubMed: 25184121]
5. Guarise A, Faccioli N, Ferrari M, Romano L, Parisi A, Falconi M. Duodenal duplication cyst causing severe pancreatitis: imaging findings and pathological correlation. World J Gastroenterol. 2006 Mar 14;12(10):1630-3. [PMC free article: PMC4124300] [PubMed: 16570360]
6. AbouZeid AA, Mohammad SA, Ibrahim SE, Fagelnor A, Zaki A. Late Diagnosis of Complete Colonic and Rectal Duplication in a Girl with an Anorectal Malformation. European J Pediatr Surg Rep. 2019 Jan;7(1):e47-e50. [PMC free article: PMC6611720] [PubMed: 31285983]

7. Khanna V, Khanna K, Srinivas M. Total midgut duplication: a ticking time bomb. BMJ Case Rep. 2018 Mar 15;2018 [PMC free article: PMC5878304] [PubMed: 29545441]
8. Soper RT. Tubular duplication of the colon and distal ileum: case report and discussion. Surgery. 1968 Jun;63(6):998-1004. [PubMed: 4869890]
9. Xiao-Ming A, Jin-Jing L, Li-Chen H, Lu-Lu H, Xiong Y, Hong-Hai Z, Nian-Yin Y. A huge completely isolated duplication cyst complicated by torsion and lined by 3 different mucosal epithelial components in an adult: A case report. Medicine (Baltimore). 2018 Nov;97(44):e13005. [PMC free article: PMC6221613] [PubMed: 30383655]
10. Rai BK, Mirza B, Hashim I, Saleem M. Varied Presentation of Congenital Segmental Dilatation of the Intestine in Neonates: Report of Three Cases. J Neonatal Surg. 2016 Oct-Dec;5(4):55. [PMC free article: PMC5117278] [PubMed: 27896163]
11. Rathi PK, Memon AS, Khatri M. Perforated ileal duplication cyst: a diagnostic dilemma. J Coll Physicians Surg Pak. 2009 Oct;19(10):665-7. [PubMed: 19811723]

Publication Details

Author Information

Authors

Sachit Anand[1]; Abdul Aleem[2].

Affiliations

[1] All India Institute of Medical Sciences

[2] Lehigh Valley Health Network, Allentown, USA

Publication History

Last Update: May 4, 2022.

Copyright

Publisher

StatPearls Publishing, Treasure Island (FL)

NLM Citation

Anand S, Aleem A. Duplication Cyst. [Updated 2022 May 4]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan-.