

Presacral Noncommunicating Enteric Duplication Cyst

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Background: Gastrointestinal (GI) tract duplication cysts or enteric duplication cysts are rare congenital malformations sometimes found on the mesenteric aspect of segments of the alimentary tract. Enteric duplication cysts are lined by normal GI epithelium and may be classified as foregut, mid-gut, and hindgut cysts. Except in very rare cases of retroperitoneal enteric duplication cysts, these cysts communicate with the GI tract and share a common blood supply. Concurrent congenital malformations are not uncommon and malignant transformation within enteric duplication cysts has also been reported.

Methods: We describe a case of a noncommunicating enteric duplication cyst in a 52-year-old woman.

Results: The patient presented with a presacral cystic mass requiring frequent drainage procedures that was primarily believed to be of neural origin. Upon resection, the lesion contained heterotopic tissue, including ciliated bronchial epithelium, squamous and transitional epithelia, and pancreatic and gastric tissue. Focal, low-grade intestinal adenoma was present, but malignancy was not detected in this case.

Conclusion: To our knowledge, this is the sixth reported case of a noncommunicating enteric duplication cyst in the English medical literature.

Introduction

An enteric duplication cyst is a cystic or tubular congenital malformation located on the mesenteric side of the gastrointestinal (GI) tract that firmly attaches to or shares the gut wall. Theories regarding the origin of GI duplications include diverticulization, canalization defects, intrauterine vascular accident, split notochord theory, and the traction hypothesis between the intestinal endoderm and the overlying structures.¹⁻⁴ Enteric duplication cyst is rare, with a prevalence of 1 out of 4,500 individuals, and it may originate from anywhere in GI tract.⁵⁻⁷

Enteric duplication cysts are lined with epithelium similar to that of the adjacent normal GI mucosa and are classified by location as foregut, midgut, or hindgut.^{5,8} In order of decreasing frequency, the most common sites of enteric duplication cysts are the ileum, stomach, and appendix, with the rarest site being the gastroduodenal region.^{9,10} Criteria to establish the diagnosis of enteric duplication cyst include the presence of a double-walled cyst, simulating the normal anatomy

of the intestine, lined with enteric-type epithelium, and sharing a common blood supply with its neighboring alimentary tract.¹¹ Rare subtypes of retroperitoneal enteric duplication cysts do not communicate with the intestine and have their own exclusive blood supply.^{2,12}

A patient's presenting symptoms depend on the anatomical location, size, and the inner lining of the cyst and may include abdominal pain, rectal bleeding, and hematuria. Enteric duplication cyst may also be incidentally discovered in asymptomatic patients. However, enteric duplication cysts become symptomatic in a person's early childhood and present in the newborn period as an abdominal mass. Few adulthood cases of enteric duplication cyst have been reported.^{8,11,13}

Intussusception, volvulus, or intestinal obstruction and, in rare cases, malignant transformation may also occur.^{8,14} In more than 50% of cases, other associated malformations such as vertebral defects and other GI duplications may be present.¹⁰ Ultrasonography, computed tomography (CT), and magnetic resonance imaging (MRI) remain the main diagnostic tools.¹⁴

Herein we present a case of enteric duplication cyst initially believed to be of neural origin. In addition, we provide a review of the literature for similar cases and discuss the diagnostic challenges presented by enteric duplication cysts.

Case Report

A 52-year-old woman presented with a history of a cystic presacral mass that was partially and initially resected via an abdominal approach in 2006. She sustained multiple recurrences, which were all managed by percu-

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taneous image-guided drainage. She was subsequently referred to our treatment team approximately 7 years after her initial diagnosis with worsening rectal pain.

Findings on both CT and pelvic MRI revealed a posterior pelvic bilobed cystic mass extending from the puborectalis posteriorly to the sacrum and coccyx and displacing the rectum anteriorly (Fig 1). The lesion also involved part of the levator muscle and had 2 cystic components measuring 3.4 and 3.2 cm, respectively. The lesion showed high-signal intensity on T1-weighted sequences and somewhat low-signal intensity on T2-weighted sequences. The lesion did not show suspicious enhancement, and the vagina and bladder were not involved.

The decision was made to proceed with surgical resection via a local, transcoccygeal Kraske approach. To allow for intraoperative identification of rectothecal communication (in the event of pseudomeningocele), preoperative lumbar puncture with intrathecal injection of fluorescein dye was performed.

During surgery, a large cystic mass to the left of the midline crossing anterior to the coccyx in a dumbbell shape was identified. The mass was adherent to the posterior rectal wall and appeared to caudally extend to the level of the external sphincter. It was completely excised by the Kraske approach. The cystic mass had no communication with the rectum or the thecal sac.

Gross examination revealed an irregular fragment of fibrous and adipose tissue measuring 7 × 6 × 3 cm and weighing 77 g. Upon sectioning the specimen, 3 cystic lesions were found, ranging from 3 to 3.5 cm in their greatest dimensions. Two of the cysts were previously opened and were connected to each other. The third cyst was independent and contained a large amount of tan-brown colored viscous material. The inner walls of the cysts were pink-tan to dusky red in color, focally erythematous and hemorrhagic, and averaged less than 0.1 cm in thickness. The fibroadipose tissue surrounding the cysts was grossly unremarkable.

Routine hematoxylin and eosin stain was done, and immunohistochemical stains were obtained with a BenchMark automated platform (Ventana Medical Systems, Tucson, Arizona) using the ultra-view DAB Detection Kit (Ventana) and antibodies cytokeratin (CK) CK7, CK20, transcription termination factor 1 (TTF1), p63, smooth muscle myosin heavy chain, and GATA binding protein 3. A trichrome special stain was also performed. Immunostain for p16 and synaptophysin were not contributory. The immunostains for p53 and Ki67 were also performed.

Results

Histopathology examination showed the cysts to be lined by intestinal-type epithelium with focal adenomatous changes surrounded by a thick muscle coat (Fig 2). The lesions demonstrated heterotopic tissue,

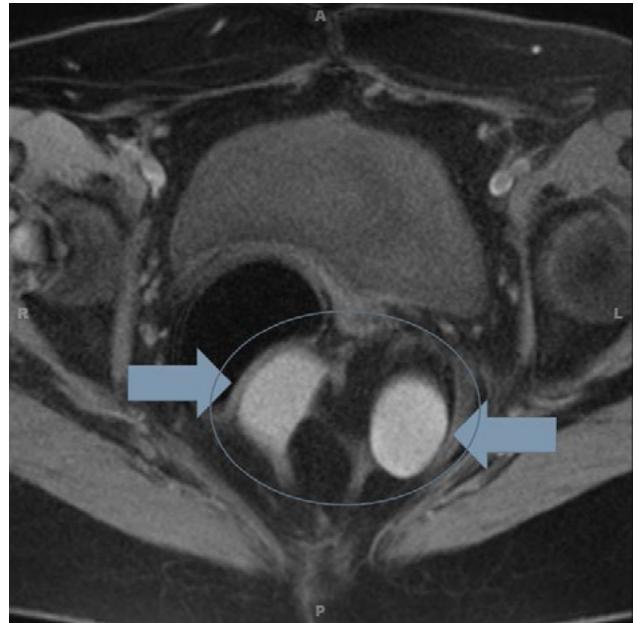


Fig 1. — Axial magnetic resonance imaging showing a presacral dumbbell shape lesion with high signal on T1-weighted images (T1 FL 2D FS).

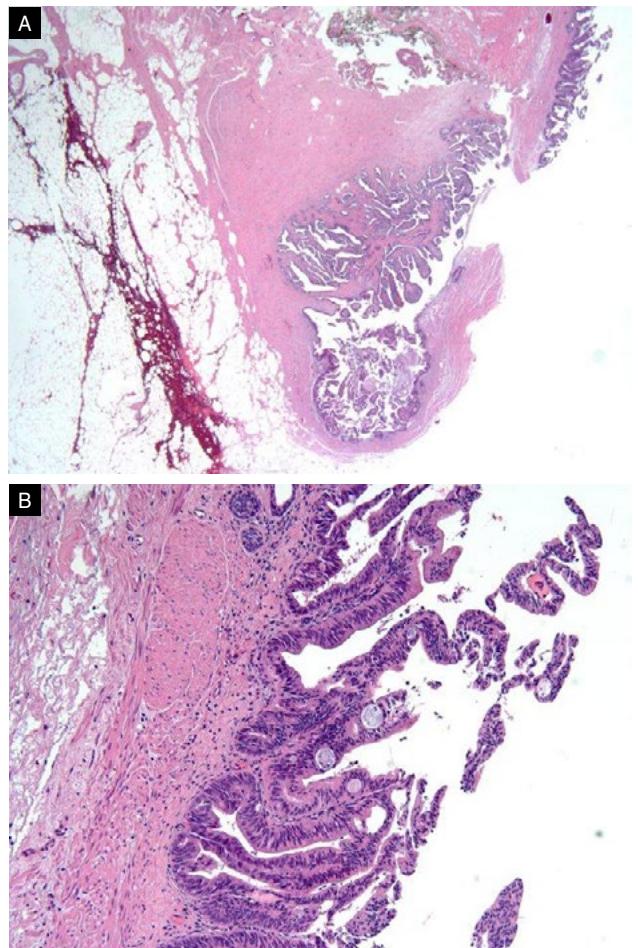


Fig 2A–B. — (A) Histology of the entire cyst at low power (H & E, × 10). (B) Component of the cyst lined by adenomatous colonic-type epithelium (H & E, × 100). H & E = hematoxylin and eosin.

including ciliated bronchial epithelium (Fig 3A), squamous and transitional epithelia (Figs 3B and 3C), and pancreatic and gastric tissue (Fig 3D).

The adenomatous epithelium and the adjacent intestinal mucosa were positive for CK7 and negative for CK20. The TTF1 immunostain was negative. The squamous and urothelial epithelia were positive for p63 (Fig 4). The inner and outer smooth-muscle layers surrounding 1 of the cysts was highlighted by smooth-muscle myosin and trichrome special stain. Ki-67 and p53 immunostains did not support high-grade dysplasia or invasive carcinoma.

Findings after trichrome special stain, which was used to highlight the layers of muscularis propria, supported the final diagnosis (Fig 5).

Discussion

Enteric duplication cysts are rare congenital lesions that can originate from anywhere in the GI tract from the oropharynx to the anus, including the retrorectal and presacral space.¹⁻⁴ The key diagnostic features for enteric duplication cysts remain the same regardless

of the location and include proximity to the GI tract, a 2-layered coat of well-developed smooth muscle, and a GI-type epithelium.^{15,16}

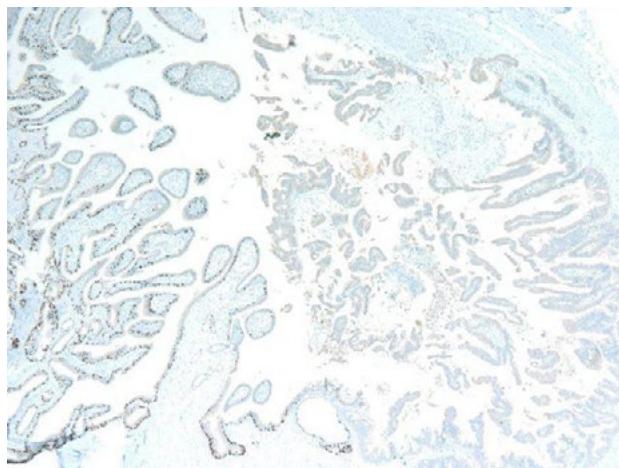


Fig 4. — p63 immunostain marking the squamous epithelium (immunohistochemistry, $\times 400$).

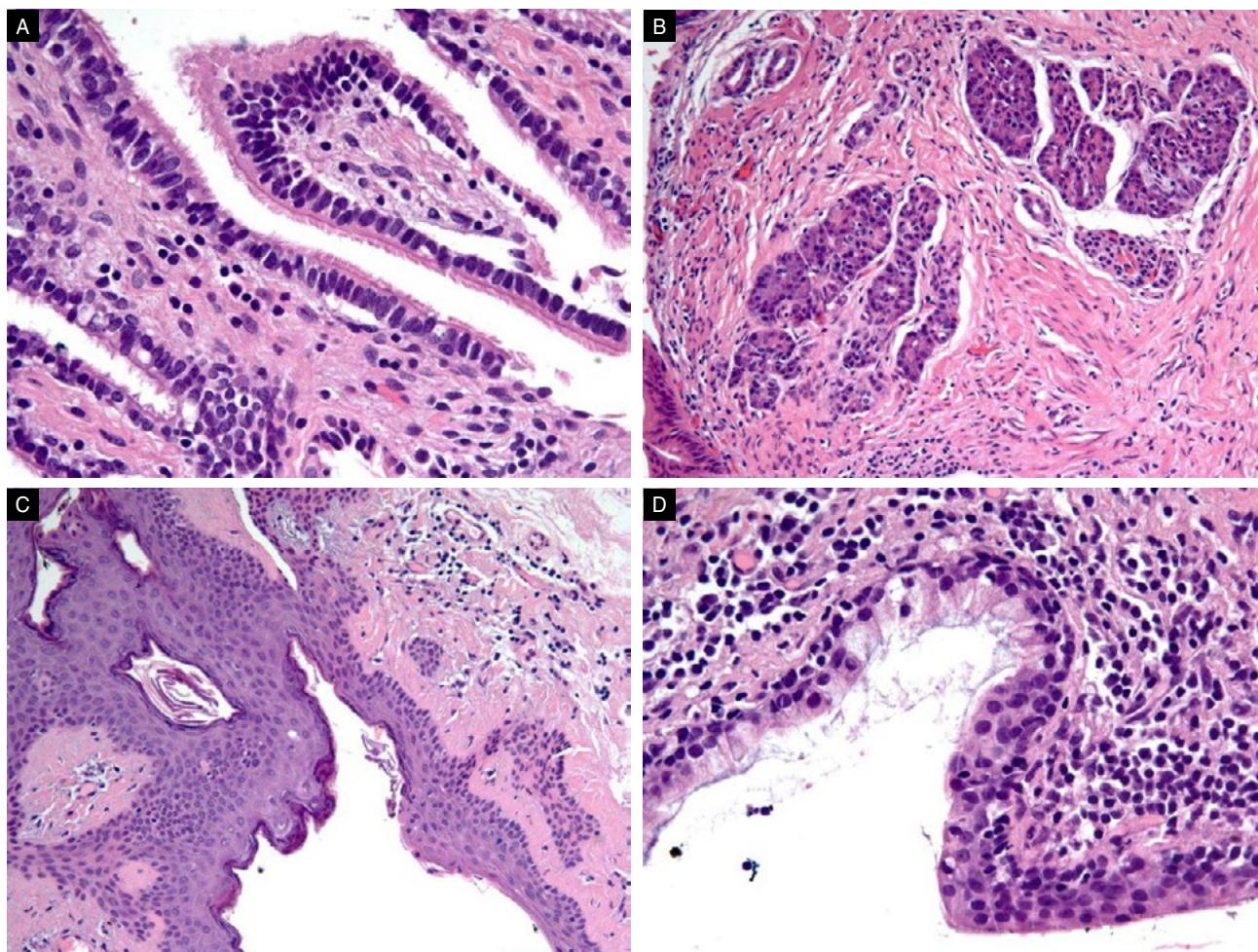


Fig 3A–D. — Heterotopic components within the cyst. (A) Ciliated epithelium (H & E, $\times 400$). (B) Squamous epithelium (H & E, $\times 200$). (C) Mucinous metaplasia (H & E, $\times 400$). (D) Pancreatic ectopia (H & E, $\times 200$). H & E = hematoxylin and eosin.

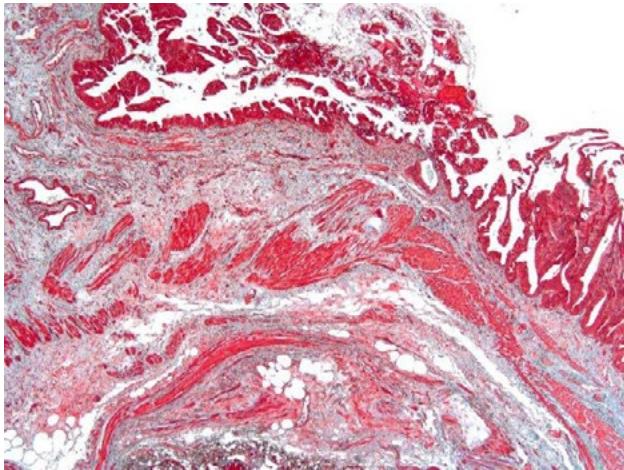


Fig 5. — Trichrome special stain highlighting the layers of muscularis propria, supporting the final diagnosis (immunohistochemistry, $\times 400$).

Rectal duplication cysts belong to the family of developmental cysts and usually present as unilocular cystic masses located in the prerectal space.¹⁷ They are lined with intestinal or respiratory epithelium and are associated with well-defined muscle layers and myenteric plexus.¹⁷

Enteric duplication cysts may have various types of presenting features.¹⁸ For example, enteric duplication cysts found in the duodenum may have communication with the biliary tract or the pancreatic duct.⁶ Those of gastric origin may involve the pancreas and may contain heterotopic tissues such as gastric mucosa, pancreatic tissue, or bronchial ciliated epithelium, or all 3 types, as was observed in our case.¹⁹⁻²¹ Correlating the radiological findings with the histopathological features is useful for making the correct diagnosis of enteric duplication cyst.

In our case, the enteric duplication cysts did not communicate with the GI tract. Such noncommunicating, isolated enteric duplication cysts have GI epithelium and a wall similar to that seen in regular enteric duplication cysts but without an anatomical association with the alimentary tract. This feature has previously been reported, but it is rare: Approximately 8 cases of this type have been reported in the English literature.²⁰ Of the cases reported, 2 were infected cases of isolated duplication cysts. One was an isolated enteric duplication cyst with features of a classic enterogenous duplication cyst, and 1 case showed mucinous cystadenoma arising within an isolated duplication cyst of the ileum.^{20,21}

Differential Diagnosis

Clinically, the differential diagnosis of a retrorectal mass is broad and includes both benign and malignant entities ranging from inflammatory, congenital or developmental cysts, neurogenic and osseous lesions like chordoma, or anterior sacral meningocele.^{16,22}

Retrorectal developmental cysts include different types of lesions such as duplication cysts, tailgut cysts, epidermoid cysts, dermoid cysts, and teratomas. Retrorectal developmental cysts are rare and may be misdiagnosed as a supralelevator abscess or a complex anal fistula.²³ A retrorectal mass with a major cystic component is highly suggestive of a developmental disorder, and the epidermoid, dermoid, and rectal duplications cysts are more often unilocular.²⁴

On CT, tailgut cysts appear as retrorectal masses with well-defined smooth borders and uneven attenuation. Calcifications and invasion of surrounding tissues are uncommon findings for tailgut cysts.²⁵ Tailgut cysts have a female predominance, whereas enteric duplication cysts are equally present in both sexes.^{15,16}

In general, duplication cysts are unilocular developmental cysts with GI- or respiratory-type epithelium. The GI epithelium reveals villi, crypts, and glands that replicate normal gut mucosa. By contrast, tailgut cysts are usually multicystic or multiloculated, lined with a variety of stratified squamous, transitional, stratified columnar, ciliated pseudostratified columnar, and gastric-type mucosa. However, when seen in conjunction with enteric duplication cysts, the mucosa usually is associated with a regular and continuous 2-layer muscle coat containing a nerve plexus. This feature is characteristic of duplication cysts and is not seen in tailgut cysts, where the smooth muscle is disorganized and focally well formed.^{17,25-30}

Cuboidal, transitional, or columnar epithelium can be seen in enterogenous cysts and tailgut cystic hamartomas. Oftentimes, they are multilocular, and disorganized smooth-muscle fibers can be identified surrounding their thin walls.²²

A developmental cyst with stratified squamous epithelium and skin appendages is suggestive of a dermoid cyst, whereas no skin appendages are seen in the epidermoid subtype. These cysts lack smooth muscle in their walls.

Teratomas are also included in the differential diagnosis of developmental retrorectal cysts. Teratomas contain heterogeneous structures from all 3 embryonic layers. Mature teratomas manifest as complex, well-circumscribed cystic and solid masses with calcifications along with epithelial lining, skin adnexa, heterologous mesenchymal tissues, and neural tissues. Primitive endoderm, mesoderm, and ectoderm are characteristic of immature teratoma. Germ cell-derived neoplastic cells are seen in malignant teratomas. Intralesional calcifications, bony destruction of the coccyx and sacrum, and hypoattenuating fat within the lesions are features of teratomas and were not seen in our case.^{22,24,27,28,31-33}

Developmental cysts are generally benign but do have the potential to undergo premalignant and malignant transformations. The presence of adenocarcinoma within a completely isolated duplication cyst

has been reported.^{34,35} These cases may manifest as bowel-like cysts with no anatomical and luminal connection with the adjacent GI tract and with a separate vascular pedicle. Development of adenocarcinomas and neuroendocrine tumors has also been reported in rectal tailgut cysts. One-half of all reported cases of presacral carcinoids are thought to have developed in a preexisting tailgut cyst or teratoma.^{29,32,34,36,37}

Conclusions

Enteric duplication cysts are rare congenital anomalies that should be considered in the differential diagnosis of a retrorectal mass. Careful review of the clinical presentation, radiological features, and gross examination of specimens and its surrounding tissue will help to narrow the diagnostic possibilities, while histopathology and immunohistochemistry are critical to establish the final diagnosis of enteric duplication cyst.

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References

- Mandhan P, Ehsan TM, Al-Sibai S, et al. Noncommunicating multiple intra-abdominal enteric duplication cysts. *Afr J Paediatr Surg.* 2014;11(3):276-278.
- Gümüş M, Kapan M, Gümüş H, et al. Unusual noncommunicating isolated enteric duplication cyst in adults. *Gastroenterol Res Pract.* 2011;2011:323919.
- Atalar MH, Cankorkmaz L, Ozer H, et al. A huge duplication cyst of the ileum. *Pol J Radiol.* 2013;78(3):70-73.
- Sheikh MA, Latif T, Shah MA, et al. Ileal duplication cyst causing recurrent abdominal pain and melena. *APSP J Case Rep.* 2010;1(1):4.
- Stern LE, Warner BW. Gastrointestinal duplications. *Semin Pediatr Surg.* 2000;9(3):135-140.
- Lopez-Fernandez S, Hernandez-Martin S, Ramirez M, et al. Pyloro-duodenal duplication cysts: Treatment of 11 cases. *Eur J Pediatr Surg.* 2013;23(4):312-316.
- Gebesce A, Korkmaz M, Keles E, et al. Importance of the ultrasonography in diagnosis of ileal duplication cyst. *Gastroenterol Res Pract.* 2013;2013:248625.
- Blank G, Königsrainer A, Sipos B, et al. Adenocarcinoma arising in a cystic duplication of the small bowel: case report and review of literature. *World J Surg Oncol.* 2012;10:55.
- Rasool N, Safdar CA, Ahmad A, et al. Enteric duplication in children: Clinical presentation and outcome. *Singapore Med J.* 2013;54(6):343-346.
- Bal HS, Kisku S, Sen S, et al. A retroperitoneal enteric duplication cyst communicating with the right upper ureter in an infant. *BMJ Case Rep.* 2014;9:2014.
- Choi SO, Park WH, Kim SP. Enteric duplications in children--an analysis of 6 cases. *J Korean Med Sci.* 1993;8(6):482-487.
- Ho YC. Total colorectal and terminal ileal duplication presenting as intussusception and intestinal obstruction. *World J Gastroenterol.* 2012;18(43):6338-6340.
- Patel MP, Meisheri IV, Waingankar VS, et al. Duplication cyst of the pylorus--a rare cause of gastric outlet obstruction in the newborn. *J Postgrad Med.* 1997;43(2):43-45.
- Deesomsak M, Aswakul P, Junyangdikul P, et al. Rare adult gastric duplication cyst mimicking a gastrointestinal stromal tumor. *World J Gastroenterol.* 2013;19(45):8445-8448.
- Liaqat N, Latif T, Khan FA, et al. Enteric duplication in children: a case series. *Afr J Paediatr Surg.* 2014;11(3):211-214.
- Joyce EA, Kavanagh DO, Winter DC. A rare cause of low back pain: Report of a tailgut cyst. *Case Rep Med.* 2012;2012:623142.
- Raisolsadat SM, Zabolinejad N, Tabrizian-Namini F, et al. Tailgut cyst in an infant with imperforate anus: a case report. *Iran J Pediatr.* 2013;23(5):597-600.
- Glasgow SC, Dietz DW. Retrorectal tumors. *Clin Colon Rectal Surg.* 2006;19(2):61-68.
- Baek SW, Kang HJ, Yoon JY, et al. Clinical study and review of

articles (Korean) about retrorectal developmental cysts in adults. *J Korean Soc Coloproctol.* 2011;27(6):303-314.

- Kim H, Kim JH, Lim JS, et al. MRI findings of rectal submucosal tumors. *Korean J Radiol.* 2011;12(4):487-498.
- Kang JW, Kim SH, Kim KW, et al. Unusual perirenal location of a tailgut cyst. *Korean J Radiol.* 2002;3(4):267-270.
- Jang SH, Jang KS, Song YS, et al. Unusual prerectal location of a tailgut cyst: a case report. *World J Gastroenterol.* 2006;12(31):5081-5083.
- Gunkova P, Martinek L, Dostalík J, et al. Laparoscopic approach to retrorectal cyst. *World J Gastroenterol.* 2008;14(42):6581-6583.
- Bathla L, Singh L, Agarwal PN. Retrorectal cystic hamartoma (tailgut cyst): report of a case and review of literature. *Indian J Surg.* 2013;75(suppl 1):204-207.
- Graadt van Roggen JF, Welvaart K, de Roos A, et al. Adenocarcinoma arising within a tailgut cyst: Clinicopathological description and follow up of an unusual case. *J Clin Pathol.* 1999;52(4):310-312.
- Cho BC, Kim NK, Lim BJ, et al. A carcinoembryonic antigen-secreting adenocarcinoma arising in tailgut cyst: clinical implications of carcinoembryonic antigen. *Yonsei Med J.* 2005;46(4):555-561.
- Alzarraa A, Mousa H, Dickens P, et al. Idiopathic benign retroperitoneal cyst: a case report. *J Med Case Rep.* 2008;2:43.
- Chhabra S, Wise S, Maloney-Patel N, et al. Adenocarcinoma associated with tail gut cyst. *J Gastrointest Oncol.* 2013;4(1):97-100.
- Govaerts K, Van Eyken P, Verswijvel G, et al. A bronchogenic cyst, presenting as a retroperitoneal cystic mass. *Rare Tumors.* 2012;4(1):e13.
- Ramakrishna HK. Intestinal duplication. *Indian J Surg.* 2008;70(6):270-273.
- Ekbote G, Pokharkar AB, Moon P. A rare case of perforated tubular ileal duplication in 72-year-old male. *Indian J Surg.* 2013;75(suppl 1):418-420.
- De Roeck A, Vervloessem D, Mattelaer C, et al. Isolated enteric duplication cyst with respiratory epithelium: case report and review of the literature. *Eur J Pediatr Surg.* 2008;18(5):337-339.
- Pins MR, Compton CC, Southern JF, et al. Ciliated enteric duplication cyst presenting as a pancreatic cystic neoplasm: report of a case with cyst fluid analysis. *Clin Chem.* 1992;38(8 pt 1):1501-1503.
- Shin SY, Cho MY, Ryu H, et al. Adenocarcinoma originating from a completely isolated duplication cyst of the mesentery in an adult. *Intest Res.* 2014;12(4):328-332.
- Okamoto T, Takamizawa S, Yokoi A, et al. Completely isolated alimentary tract duplication in a neonate. *Pediatr Surg Int.* 2008;24(10):1145-1147.
- Spada F, Pelosi G, Squadroni M, et al. Neuroendocrine tumour arising inside a retro-rectal tailgut cyst: Report of two cases and a review of the literature. *Ecancermedicalscience.* 2011;5:201.
- Liang JJ, Alrawi S, Fuller GN, et al. Carcinoid tumors arising in tailgut cysts may be associated with estrogen receptor status: case report and review of the literature. *Int J Clin Exp Pathol.* 2008;1(6):539-543.