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Adenocarcinoma arising in a rectal duplication cyst with distant metastasis. A case report and a review of the recent literature

INTRODUCTION: *Rectal duplication cysts are rare cystic lesions, arising from the hindgut and classified as congenital/developmental tumors of the presacral space. Their clinical presentation is nonspecific, the diagnosis remains difficult and their management is aided by a multidisciplinary evaluation.*
CASE REPORT: *We report the case of a 55-year-old woman with a cystic mass located in the retrorectal space and identified incidentally on a CT scan. Following imaging studies, surgical resection by a posterior approach (Kraske procedure) was carried out and an adenocarcinoma arising in a duplication cyst of the rectum was present an uncommon case of a rectal duplication cyst with malignant transformation and distant metastasis, describe the clinical, radiologic and pathologic findings and discuss tidentified by microscopy.*
CONCLUSION: *We phe embryological basis of rectal duplication cysts and the surgical anatomy of the presacral space.*

KEY WORDS: Rectal adenocarcinoma, rectal duplication cyst, Retrorectal space

Introduction

Rectal duplication cysts are rare congenital cystic lesions arising from the embryologic hindgut[1]. These lesions represent only 5% of the alimentary tract duplications[2,3]. In the only 28 cases with rectal duplication cyst in adults existing literature have been presented up to now. Six of them described adenocarcinoma that had been developed to the cystic lesion. The differential diagnosis includes other congenital or developmental cysts occurring in the retrorectal (presacral) space (tailgut cysts, dermoid and epidermoid cysts, teratoma), neurogenic neoplasmas (anterior sacral meningocele, sacrococcygeal chordomas), inflammatory or metastatic lesions, osseus and miscellaneous tumors [1,4,5,6]. Although the majority of lesions remain asymptomatic [1] and the diagnosis presents significant difficult [3], the patients in most of the cases are presenting with rectal, low back, suprapubic or sciatic pain [2], intestinal obstruction, local bleeding, urinary or fecal incontinence, fistulation, infection and sexual dysfunction [1,4,7].
The clinical presentation is associated with the size, the invasion of the tumor to the adjacent tissues, the presence of fistulation, infection, ectopic gastric mucosa and malignant transformation [2]. A typical diagnostic approach include clinical examination, computed tomography (CT) and conventional or endorectal magnetic resonance imaging (MRI). Transanal ultrasonography and proctoscopy may also be required in order to confirm the diagnosis

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[1,2,4,6,7]. Either CT or MRI have become the "gold" standard for the preoperative evaluation of the presacral tumors [8,9]. These imaging studies are helpful, but diagnosis should always be confirmed by histology.[5] Malignant transformation is extremely rare and was first reported in a 38-year-old female's rectum in 1932 [10]. Complete surgical excision is indicated to establish the diagnosis and prevent complications like sepsis, bleeding or malignant change. A transanal, posterior or a combined abdominoperineal surgical approach can be performed depending on the position of the cyst and to its relationship with the surrounding structures. Generally small lesions below the level of S3 can be removed through a posterior approach. If the upper extension of the tumor is above the level of S3 or the dimensions of the mass are extended, a combined anterioposterior approach should be prefered [4,11].

Case Report

A 55-year old woman evaluated for acute abdominal pain and the CT scan revealed acute ruptured appendicitis which was successfully treated with open appendicectomy in another hospital. The abdominopelvic CT scan revealed also an asymptomatic cystic lesion located in the presacral space, measuring approximately 6,8 x 4,3 cm. Upon further investigation one year later with MRI scan confirmed the diagnosis of a presacral cystic mass as shown in figures 1-2. In 2016 – three years after diagnosis - the patient presented for the first time to our surgical department for further investigation and management of the mass. The rest medical history of the patient was unremarkable but she had reported a feeling of heaviness in the pelvis and low back pain during the last years. She underwent a CT-guided percutaneous drainage and the cytology test of the sample did not reveal evidence of malignancy. Endoscopic examination

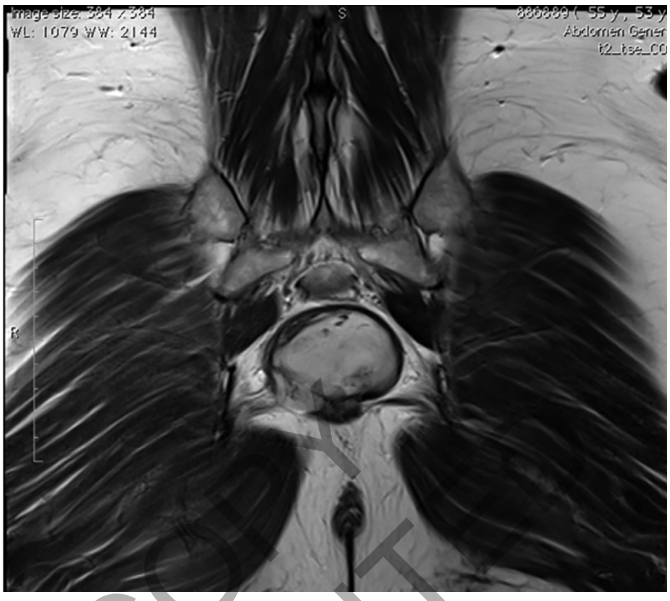

Fig. 2: Preoperative MRI scan (T2).

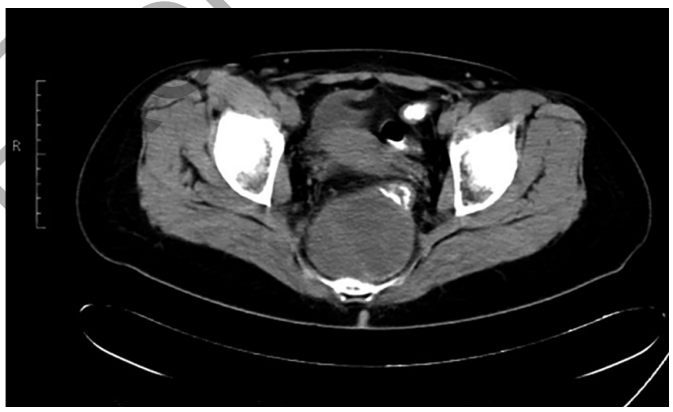

Fig. 3: Preoperative CT scan.

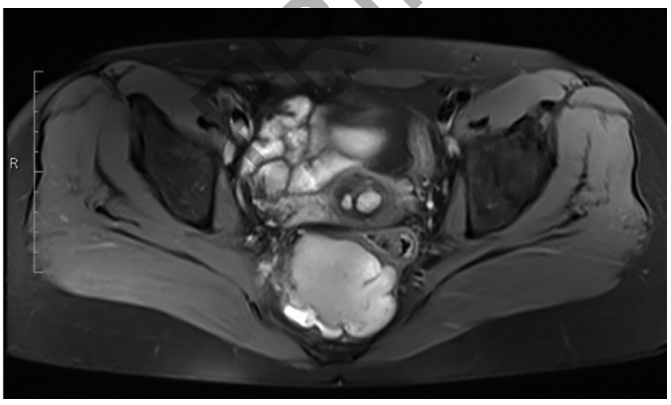

Fig. 1: Preoperative MRI scan (T1).

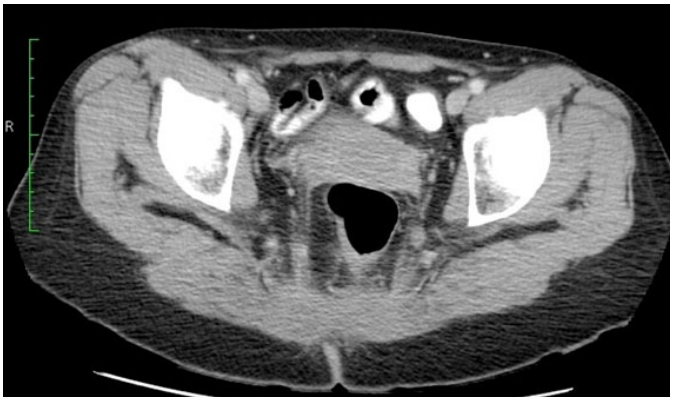

Fig. 4: Postoperative CT scan.

with gastroscopy and colonoscopy some months ago came up without abnormal findings. A repeat CT scan revealed a large oval hypodense cystic lesion arising from the presacral region, measuring 77,8 x 66,2mm with smooth well-circumscribed margins and lobular appearance, separated with septations, located posterior to the rectum, pushing the sigmoid colon and abutting the pelvic surface of the sacrum without evidence of penetration to either of them (Fig. 3). The decision was to attempt radical surgery and following the image findings, a posterior transcoccygeal excision of the tumor was carried out. The mass was dissected free from surrounding structures and it was completely resected en block with the coccyx. The histopathological examination of the sample confirmed a cystic lesion composed from inside inward by columnar epithelium, hypertrophic muscular layer with circular smooth bundles, a few ganglion cells and neurous plexus lying between the bundles, similar to the intestinal wall and serosa. The columnar epithelium presented with dysplastic features and a development of a tubular adenocarcinoma was revealed, characterized by increased production of extracellular mucus and with penetration of all the layers of the cystic wall. Immunohistochemically, the neoplastic cells were positive for CK20, CDX2 and CK7. The final diagnosis was a mucinus adenocarcinoma arising from a rectal duplication cyst with extended penetration of the coccyx. She was admitted for postoperative chest and abdominal CT scan (Fig. 4) and pulmonary metastatic lesions were observed.

Discussion

Retrorectal region, according to Jackman et al., is not a space but becomes an appartment only when the rectum is deplaced anteriorly by a mass [12]. The retrorectal space is a pelvic continuation of the retroperitoneum [8], bounded anteriorly by the fascia propria of the rectum at about the junction of the second and third sacral segments. Fascia propria of the rectum (or mesorectal visceral fascia) is a distinct layer of connective tissue. It is considers as an extension of the pelvic fascia that limits the mesorectum posteriorly and lies anterior to the presacral fascia separated the presacral space. It is present mainly in the lateral and posterior extraperitoneal portion of the rectum [13]. Posteriorly, that space is bounded by the presacral fascia, which is a thickened and strong part of the parietal pelvic fascia that covers the concavity of sacrum and coccyx. The space is also bounded inferiorly by the rectosacral fascia (or Waldeyer's fascia), which is a thick fascial reflection that passes from S2, S3, or S4 to the rectal fascia propria, inserting approximately 3 to 5cm above the anorectal junction. Below Waldeyer's fascia lays the supralevator space. The retrorectal region extends cranially to the peritoneal reflection, as well as the lateral boundaries are again the peritoneal reflection assisted by the iliac arteries, the ureters and lateral rectal stalks and ligaments. The presacral space normally contains fat, loose areolar and connective tissue, the middle sacral artery and presacral veins covered by the Waldeyer's fascia, the superior and middle rectal vessels, the internal iliac vessels, lymphatics, and sacral branches of the sympathetic and parasympathetic plexus of nerves [1,4,14-16].

As the retrorectal region is the site of fusion of the developing hindgut [4], rectal duplication cysts can be presented here with the first case reported in 1885 [17]. These tumors are rare developmental cystic lesions and they are diagnosed based on three histological criteria as defined by Ladd and Gross[18]: (1) continuity or contiguity with the rectum, (2) well defined muscular wall with a myenteric plexus, and (3) mucosal lining [2,7]. They usually appeared multilobular and a variety of epithelial linings can be seen, including columnar, transitional, squamous and cuboidal epithelia. Rectal duplications can be located anteriorly to the rectum or more often posteriorly and classified as cystic (type I) and tubular (type II) duplications [2].

Many theories have been suggested to explain the development of enterogenous cysts. Veeneklass's theory [19] proposes that the duplications result from a disorder during separation of the notochord due to defective detachment of the notochord from the endoderm [2,19]. According to another hypothesis, enteric cysts are developed by persistent enteric diverticula during embryonic development [3,20-22].

Bishop and Koop [23] suggest that duplications in the abdomen usually lie within the leaves of the mesentery of the functioning intestinal tract. The mesenteric blood vessels are intimately attached to the duplication and surround it as they fan out to supply the normal intcstina1 tract beyond it [23]. The main artery that gives arterial supply to the hindgut is the inferior mesenteric artery (IMA) while the principal arterial supply to the upper two-thirds of the rectum is via the superior rectal artery which consists the pelvic continuation of the IMA and descends to the posterior wall of the upper rectum in the midline[14,15].

A possible explanation why a rectal duplication cyst - as it presented in our patient - is expected to appear more often posterior to the rectum, is based on the above theories for the development of rectal duplications, the theory of the common blood supply shared between the duplication and the native bowel and the dorsal route of the inferior mesenteric artery in order to approach posteriorly and to vascularize the hindgut during the embryogenesis.

The anatomy of the presacral space, as it mentioned above, is also useful for the surgeon during a posterior

transcoccygeal excision of a rectal duplication cyst as we can sacrifice structures with safety if it is necessary for the progress of the operation. To make our suggestion more clear, we refer a relative example: The lower pair of sacral spinal nerves S5 which passes laterally through the sacral hiatus, immediately above the level of sacral cornua, as well as the coccygeal nerves Co which pass laterally above the level of coccygeal cornua, penetrate each of them the coccygeus muscle, overlying the sacrotuberous and sacrospinous ligaments and they pass superficially in order to innervate the perianal skin. The nerves of coccygeal plexus (S5, Co) can be sacrificed during the operation without failure of motor (muscular) function. The sacral nerves S4 can also be sacrificed without affecting to the anorectal function, as the inferior branches of the pudendal nerve carry fibers from S2 to S4 levels innervating the skin and the skeletal muscles of the perineum, including the external anal sphincter. [24]

Riassunto

Le cisti di duplicazione rettale sono rare lesioni cistiche, derivanti dall'hindgut e classificate come tumori congeniti / sviluppatisi dello spazio presacrale. La loro presentazione clinica non è specifica, la diagnosi rimane difficile e la loro gestione è fondata su una valutazione multidisciplinare. Riportiamo il caso di una donna di 55 anni con una massa cistica situata nello spazio retrorettale e identificata incidentalmente su una scansione CT. Dopo studi di imaging, è stata effettuata la resezione chirurgica da un approccio posteriore (procedura di Kraske) ed è stata identificato istologicamente un adenocarcinoma insorto in una cisti di duplicazione del retto. Si tratta dunque di un caso raro di una cisti di duplicazione rettale con trasformazione maligna e metastasi a distanza, e vengono descritte le indagini, radiologiche e patologiche e discutessa la base embriologica delle cisti di duplicazione rettale e l'anatomia chirurgica dello spazio presacrale.

References

1. Grundfest-Broniatowski S, Krishnaney A: *Diagnosis and Management of Sacral and Retrorectal Tumors*. In: Fazio VW, Church JM, Delaney CP, Kiran RP. *Current therapy* in *colon and rectal surgery*. Philadelphia, PA: Elsevier; 2017; 121-30.

2. Gupta DK, Sharma S: *Rectal duplication and anal canal duplication. In:* Holschneider AM, Hutson JM: *Anorectal malformations in children: Embryology, diagnosis, surgical treatment, follow-up*. Berlin: Springer; 2006; 231-36.

3. Parvaiz A, Stevens RJ, Lamparelli MJ, et al.: *A rare case of adenocarcinoma arising within a duplication cyst of the rectum: Curative excision with 9-year follow-up*. Ann R Coll Surg Engl, 2005; 87: W8-W10.

4. Neale J: *Retrorectal Tumors.* Clinics in Colon and Rectal Surgery, 2011; 24(03):149-60.

5. Kim JH, Jin SY, Hong SS, Lee TH: A *carcinoid tumour arising within a tailgut cyst: A diagnostic challenge*. Scott Med J, 2014; 59(1):e14-7.

6. Wolpert A, Beer-Gabel M, Lifschitz O, Zbar AP: *The management of presacral masses in the adult*. Tech Coloproctol, 2002; 6:43-9.

7. Hassan I, Wietfeldt D: *Presacral tumors: Diagnosis and management*. Clin Colon Rectal Surg, 2009; 22:84-93.

8. Glasgow S, Dietz D: *Retrorectal Tumors.* Clinics in Colon and Rectal Surgery, 2006; 19(2):061-68.

9. Jackson KL, Peche WJ, Rollins MD: *An unusual presentation of a rectal duplication cyst*. International Journal of Surgery Case, 2012; 3(7):314-15.

10. Ballantyne EN: *Sacrococcygeal tumours*. Arch Pathol, 1932; 14: 1.

11. Young-Fadok TM, Dozois EJ: *Retrorectal Tumors*. In: Zuidema GD, Yeo CJ,(rds): *Shackelford's surgery of the alimentary tract*. 6th ed. Philadelphia: W.B. Saunders; 2007; 2307.

12. Jackman RJ, Clark PL, Smith ND: *Rectorectal tumors*. JAMA, 1951; 145(13):956-62.

13. Garza A, Beart RW, Jr.: *Anatomy and embryology of the anus, rectum, and colon*. In: Corman ML, Corman ML: *Corman's colon and rectal surgery*. Philadelphia: Wolters Kluwer Health/Lippincott Williams & Wilkins; 2013, 1-9.

14. Skandalakis JE, Colborn GL, Weidman TA, et al.: *Skandalakis' Surgical anatomy.* 1st ed. Athens, Greece: PMP; 2004. Chapter 18

15. Lunniss PJ: *Large intestine.* In: Standring S.: *Gray's anatomy: the anatomical basis of clinical practice.* Philadelphia: Elsevier Limited; 2016; 1146-152.

16. Boutros M, Gordon PH: *Anatomy and physiology of the colon, rectum, and anal canal.* In: Fazio VW, Church JM, Delaney CP, Kiran RP: *Current therapy in colon and rectal surgery*. Philadelphia, PA: Elsevier; 2017; 3-6.

17. Middeldorf K : *Zur kanntniss der ange Bornen Sacral gasch Ulsle*. Virchows Arch [A],1885; 101:37-44.

18. Ladd, WE, Gross RE: *Surgical treatment of duplication of the alimentary tract: Enterogenous cysts, enteric cysts, or ileum duplex*. Surg Gynecol Obstet, 1940; 70:295-307.

19. Jewell CT, Miller I, Ehrlich FE: *Rectal duplications: an unusual cause of an abdominal mass.* Surgery, 1973; 74:783-85.

20. Ildstad ST, Tollerud DJ, Weiss RG, Ryan DP, McGowan MA, Martin LW: *Duplications of the alimentary tract. Clinical characteristics, preferred treatment, and associated malformations*. Ann Surg, 1988; 208:184-89.

21. O'Rahilly R, Müller F: *Human Embryology and Teratology*. 2nd ed, New York, John Wiley & Sons;1996.

22. Michael D, Cohen CR, Northover, JM: *Adenocarcinoma within a rectal duplication cyst: Case report and literature review*. Ann R Coll Surg Engl, 1999; 81:205-06.

23. Bishop HC, Koop CE: *Surgical management of duplications of the alimentary tract*. Am J Surg, 1964;107:434-42.

24. Scott-Conner CEH, Dawson DL: *Operative anatomy*. Philadelphia: Wolters Kluwer Health/Lippincott Williams & Wilkins; 2009; 714-718.