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Retrorectal multilocular cyst in a adult female

Case report and review of literature

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Retrorectal multilocular cyst in a adult female. Case report and review of the literature

Tumors occurring in the retrorectal space are heterogeneous and uncommon. Multiple embryologie remnants derived from a variety of tissues are found in this space and this explains the heterogeneous pathology of cysts and tumors that arise in retrorectal space. Presacral cysts are rare in adults. Most of the lesions are benign. The cysts may be unilocular or multilocular. They are often asymptomatic. These tumors pose a diagnostic and therapeutic interest. Presacral epidermoid cysts are classified as a type of developmental cysts. We report the case of a 57-year-old woman who presented with a painless mass posterior of the anus. Imaging studies showed a multilocular cyst (65 × 100 × 60 mm) in the retrorectal space. The patient underwent surgery via a posterior approach. Histological study revealed a epidermoid cyst. A review of literature of this rare tumor is performed.

KEY WORDS: Presacral cyst, Retrorectal epidermal cyst, Retrorectal tumours

Introduction

Retrorectal or presacral cystic tumors are rare pelvic tumors; however, most general surgeons can expect to encounter at least one patient with a retrorectal tumor during the course of their careers [1]. The retrorectal space is bounded by the sacrum posteriorly, the rectum anteriorly, the peritoneal reflection superiorly, the levator musculature inferiorly, and the ureters on each side. A variety of congenital, neurogenic, osseous, inflammatory, and miscellaneous disorders may occur within this space [1]. Developmental cysts, which account for 60% of all congenital presacral tumors, can arise from any embryonic layer. Based on cell layer of origin, developmental cysts can be epidermoid, dermoid, tailgut, or teratomas. Epidermoid cysts are relatively rare [2-4]. Most cases reported in the literature occurred in women and were largely asymptomatic [3,4]. Reports indicate that these cysts are rare in adults [2,4,5], tipically benign unilocular lesions [6,7] and often found around the time of childbirth or during gynecologic examination and evaluation [3,4]. The correct diagnosis of tumours in the retrorectal space is known to be difficult [2], despite recent advances in diagnostic modalities; ideal treatment still remains unclear. Depending on the location and /or size of the tumor as well as its relationship with surrounding tissues, various surgical approaches can be selected [8]. We describe here a rare case of multilobulated retrorectal epidermal cyst in a 57-year-old woman and discuss the incidence, preoperative diagnosis and treatment of retrorectal cystic tumors.

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Case report

A 57-year-old woman was referred to our institution for the further examination of a presacral mass. Clinical examination revealed abnormalities in the perianal area: a painless mass posterior of the anus. Digital rectal examination revealed a non-tender, extraluminal well-defined mass in the presacral region causing extrinsic compression. The overlying rectal mucosa was smooth and mobile and the upper limit of the lesion was reached. Magnetic resonant imaging confirmed a lesion mixed (solid and cystic) with multiple cysts in the presacral region, with antero-posterior, cranio-caudal and transversal maximum diameters measuring 65 × 100 × 60 mm and without clear signs of invasion of any surrounding organs. The lesion extended below S4 and one of these cysts (the largest) of approximately 5.5 x 3.5 cm is wedged in the pelvic floor muscles becoming subcutaneously (Fig. 1/a-b). The laboratory data were all within the normal range, including tumor markers. Preoperatively, the differential diagnosis included almost all presacral cystic masses. The patient underwent surgery via a posterior approach in lithotomy position, under spinal anesthesia. It was performed a curved incision concave upward in the ano-coccygeal raphe. During the operation, the elastic soft mass was palpated after subcutaneous fat excision and was adhered to the external anal sphincter muscle. The blunt dissection of mass was continued into the supra-elevator space where there was a clear cleavage plane with the rectum wall, well visible

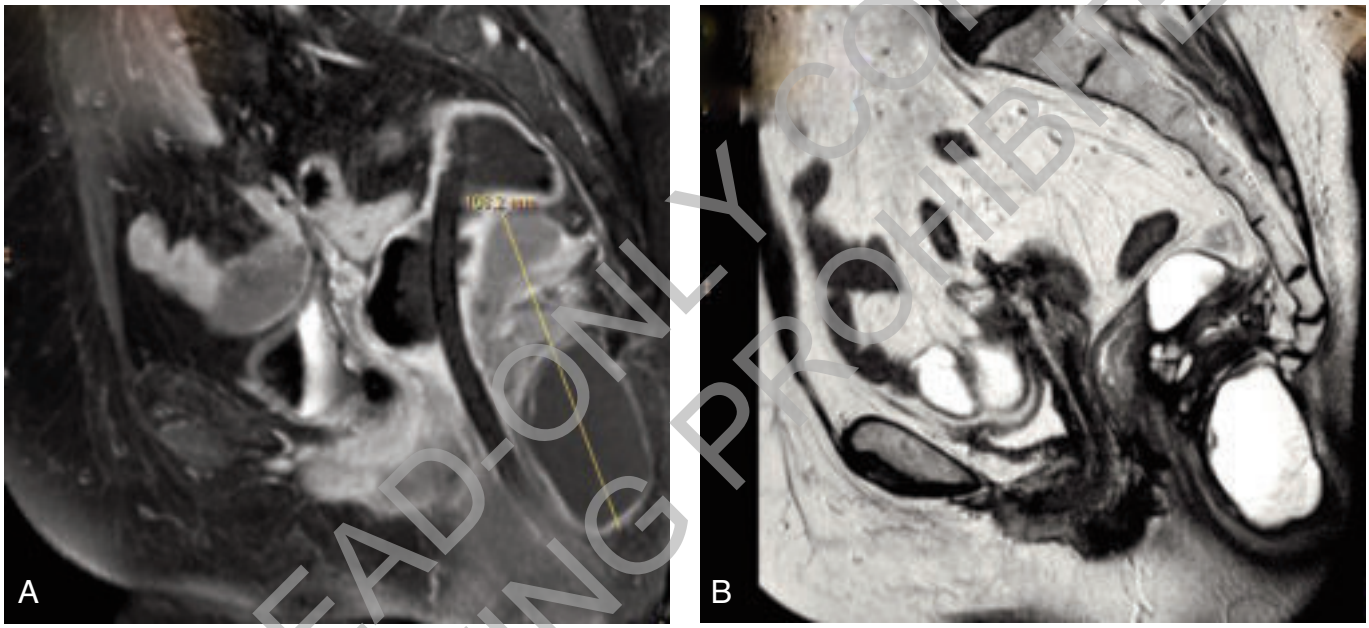

Fig. 1: (A) T1- and T2-weighted (B) magnetic resonance images. T1-weighted sagittal MRI scan showing the retrorectal multiloculated cyst and the rectum (rectal tube).

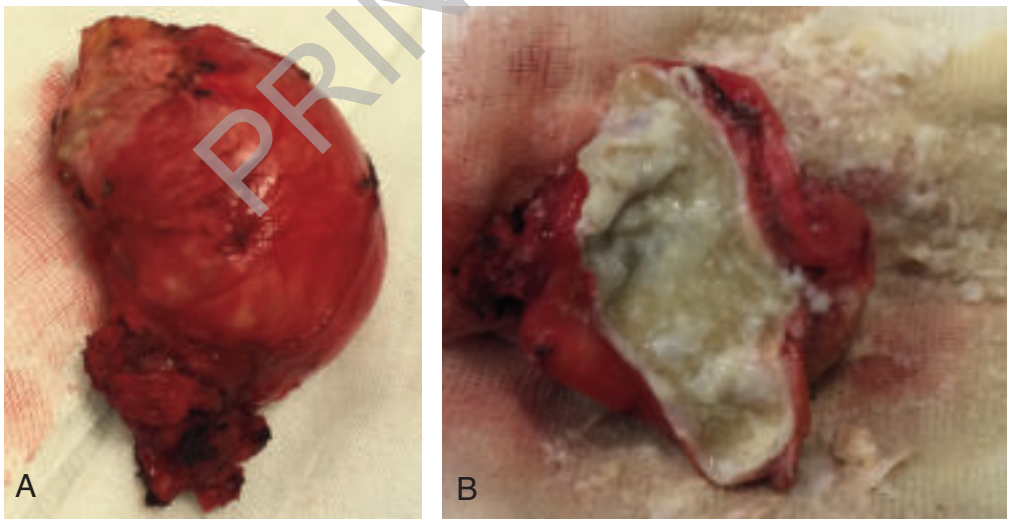

Fig. 2: (A-B) Gross examination.

superiorly. The lesion was excised without difficulty and the levator muscles was sutured with absorbable stitches; the wound was closed without placement of drain. The specimen was represented by a cystic tumor, approximately 6 x 10 cm in diameter, containing cheese-like material, but no hair or tooth matter; there was no solid component in the cystic lumen (Fig. 2/a-b). Histopathological examination classified the tumour as epidermoid cyst. The patient had an uneventful postoperative course and was discharged on the 3th postoperative day. On 6-month follow-up, the patient was doing well and was symptom-free [6].

Discussion

The incidence of retrorectal tumors in the general population is unknown, because many are asymptomatic. Uhlig and Johnson [9] demonstrated the incidence of two pre-sacral tumors per year in a metropolitan population at a nontertiary referral center and Jao et al found that retrorectal tumors accounted for 1 in 40,000 hospital admissions [10]. Only small series exist in the literature, the largest being one with 120 patients published by the Mayo Clinic in 1985 [10,11]. The classification by Uhlig and Johnson is used to classify retrorectal tumors in five categories: congenital, neurogenic, osseous, inflammatory and miscellaneous [9]. About 55-70% of all presacral tumors are congenital lesions[1]; these lesions are thought to arise from the remenants of embryonic tissue and can present as either cystic or solid lesions. Cystic congenital lesions include developmental cysts and anterior meningoceles. The solid congenital lesions consist of teratomas, sacrococcygeal chordomas, and adrenal rest tumors [12]. Developmental cysts are the most common congenital lesions; they are found only rarely in the retrorectal region of adults and these occur mostly in middle-aged women. Depending on the cell layer of origin, they can be divided into the following types: epidermoid cysts, dermoid cysts, enterogeneous cysts, tailgut cyst and teratomas [13]. Dermoid/epidermoid cysts are rare [2]. Epidermoid cyst in the presacral space was first described by Birkett [4,14] and to our knowledge, the incidence of 5-22% of all retrorectal tumors (mean 13%, Table I) [15-19] stated in recent reports appears greater than that previously reported [4]; this can be linked to a better characterization of the retrorectal masses; inflammatory lesions are not mentioned in several series [1,18,19]. Of retrorectal developmental cysts, 32,5% are epidermoid cysts in a series of 40 cases from Korea [20]. Epidermoid and dermoid cysts result from a closure defect of the ectodermal tube and thus have a squamous epithelial lining; they are tipically benign unilocular lesions; skin appendages are absent in epidermoid cyst [6]. Tail gut cysts (cystic hamartomas) are multinodular, uncapsulated, and usually well-circumscribed cysts; they are derived from tailgut remnants, the precursor of the gastrointestnal system [12]. Teratomas arise from totipotential cells that can give rise to any tissue type [1]; they can be solid or cyst and often contain both component. Teratomas are more frequently found in the pediatric population; in the adult population there is a risk of 40 to 50% malignant degeneration [12]; they tend to adhere firmly to the coccyx. Duplication cyst (enterogeneous) are thougt to develop due to sequestration of the developing hindgut [13]; these tumors usually have a multilobular apperance with multiple satellite lesions and one dominant lesion [9]. Although generally benign, malignant degeneration has been reported. Most benign cystic lesions are asymptomatic and are discovered on routine rectal examination. Although mostly asymptomatic, patients may present with symptoms resulting from local mass effect (dull pain in perianal area, constipation, rectal fullness, lower abdominal pain, lower back pain, straining, overflow incontinence, dysuria, dystocia in young women and sometimes sciatic-type pain) or with a complication. Our patient complained of a swelling posterior anus, visible inspection of the perineum. The most important complications of these cysts are infection with secondary fistulization and malignant degeneration. Epidermoid cysts may communicate with the skin creating a postanal dimple and have a high rate of infection (up to 30%); infected cysts can be easily mistaken for perirectal abscess, pilonidal disease, or fistulae in ano [21]. Malignant transformation of developmental cysts is rare. Abel reported an incidence of 8% in developmental cysts [5,22]. In Japan, on 58 cases of epidermoid cysts, have been reported 4 cases of malignant transformation; all were squamous cell carcinoma [5]. Malignant tumors that invade the sacral plexus or nerve roots can

TABLE I - *Incidence of epidermal cyst in retrorectal tumours*

Author/year	Institution	Incidence/(%)	Length study
Wang JY /1995[15]	Chang Gung Memorial Hospital	10/45(22)	1978-1992
Glasgow Sean C. /2005[16]	Washington University	5/34(14)	1981-2003
LI Guo-dong /2011[17]	Tongji University School of Medicine	6/33(18)	1998-2009
Macafee D.A.L. /2012[18]	John Goligher Colorectal Unit	3/56(5)	2002-2010
Craig A. Messick /2013[19]	Cleveland Clinic	10/87(11)	1981-2011
Totale		34/255(13)	

lead to bowel and bladder incontinence or urinary retention. The diagnosis of retrorectal cyst should be suspected in any patient with a history of repeated attempts at drainage for a fistula in ano or perirectal abscess, especially when no obvious internal rectal opening can be identified or mucoid material (rather than pus) is drained. Singer et al. reported that all of their patients with retrorectal cyst had been treated for fistula in ano, pilonidal disease, perianal abscess, postpartum pain, and psychogenic pain, before the correct diagnosis was made [23]. Physical examination focuses primarily on the perineum and rectum. Visual inspection of the perineum and perianal region can detect the existence of a posterior anal funnel-shaped dimple (fovea coccigea); a finding highly suggestive for the presence of a developmental cyst. Usually a soft prominence with a smooth intact overlying mucosa, can be felt in retrorectal area; the patient might complain of tenderness, and a feeling of pushing the liquid-fulled cyst might exist [20]. A neurological examination must be performed focusing on the lumbar and sacral roots to ascertain their involvement [11]. Plain radiographs of the pelvis are tipically normal. The exceptions are malignant tumors that may produce bone destruction of the sacrum, as well as the rare benign bone-based lesions. With endoscopy, prominent mucosa in retrorectal area can be seen if lesion is large enough, and the proximal extent of the lesion can be confirmed as well. Tomographic imaging with either CT or MRI, has become the standard for the preoperative evaluation of retrorectal tumors. A CT scan can be used to determine whether a lesion is solid or cystic, evaluate cortical bone destruction, and assess involvement of adjacent viscera [13]. On MRI, presacral cyst usually has low signal intensity on T1- weighted images and high signal intensity on T2-weighted images. However, it may have high signal intensity on T1-weighted images due to presence of mucinous materials, high protein content, or hemorrhage in the cyst [7]. Epidermoid cysts or dermoid cysts appeared unilocular, and tailgut cysts appeared as a large cyst accompanied by small peripheral cysts [7]. MRI may diagnose the tumour as benign, malignant or uncertain, but was not helpful in assessing recurrent benign tumours; cystic lesions with a smooth wall on MRI are typically benign, whereas heterogeneous tumours are usually (but not always) malignant [7,24]. However, the differentiation between presacral masses is not likely to be done purely on radiographic evidence; there is no pathogenomic finding for epidermoid cyst [3]. In our patient, the image of a multilocular lesion on MRI not deposed for an epidermoid cyst; in addition, the development of the mass outside the presacral space, becoming subcutaneously in the posterior perineum, is not reported previously in a series of case reports [2-6,8,25-28]. Other diagnostic modalities that can help in the diagnosis and management of presacral lesions include fistulograms and endorectal ultra-sound. The role of biopsy is a critical, and often misunderstood, aspect of evaluating retrorectal lesions. For resectable lesions, surgical resection is the best diagnostic and therapeutic option [21]. Biopsy also can infect previously sterile cystic lesions and lead to dissemination of the tumor cells when a retrorectal cyst is neoplastic in nature [1,16,21]. Therefore, a biopsy should only be performed if the lesion appears to be unresectable [16] and tissue diagnosis is required to guide adjuvant therapy [1,21]. The indications for biopsy can be limited to patients whose mass may represent metastatic disease or lymphoma [24].

However, Merchea et al. has recently reported the absence of recurrences at the biopsy site/tract in a series of patients undergoing preoperative biopsy; the author feel strongly that given the potential for alteration in either preoperative therapy or intraoperative tecnique, and because of limitations of imaging to make a definitive diagnosis, percutaneous biopsy should be obtained to facilitate these decisions [29]. Once a diagnosis of presacral cystic tumor is established, operative intervention is recommended, even if it is asymptomatic. The lesion may be malignant or may become malignant, like teratomas; cystic lesions can become infected spontaneously and there exists the possibility of dystocia in young women [11]. The operative approach to presacral cystic tumors will depend on the type of lesion, the extent of the lesion, and what structures are involved. Curative resection requires complete excision of the tumour, with an intact capsule for clinically benign well circumscribed lesions and *en bloc* resection with microscopically clear resection margins for malignant tumours. The common approaches for resection of retrorectal tumors are the anterior (transabdominal), the combined abdominoperineal, and the posterior (perineal) approaches [12]. The anterior approach is performed for high lesions without sacral involvement, with the lowest portion of the lesion above the level of S4. If the tumour is positioned below the mid-body of S3, a perineal approach can be considered. When the examiner's finger can palpate the upper edge of the tumor, removing the cyst by only using a posterior approach should be possible. When the tumour was larger than 10 cm, or there was difficulty with access, a coccygectomy, or excision of S5 with or without the S4 vertebrae, should be performed; this permitted good exposure [24]. Larger tumors or those extending both proximal and distal to the fourth sacral element are excised *via* a combined abdominosacral approach [1]. Minimally invasive techniques can be applied to the excision of retrorectal tumours that were previously approached via laparotomy. In the series of Fong et al., the indication for laparoscopic excision included tumours above the S3/4 junction that could not be treated with a perineal approach with benign characteristics on MRI and less than 20 cm in maximum diameter [30]. Also, transrectal excision using TEM is reported feasible and safe for a benign retrorectal lesion [31]. The prognosis depends on the type of tumour and completeness of resection. In benign tumours, survival is 100% and if the resection is complete recurrences are rare, although

they have been reported in up to 15% of developing cysts. It is therefore important to avoid excessive manipulation or rupture of the tumour [11].

Riassunto

I tumori che occorrono nello spazio retrorettale sono eterogenei e non comuni. Molteplici resti embriologici derivati da una varietà di tessuti, si trovano in questo spazio e questo spiega la patologia eterogenea di cisti e tumori che insorgono nello spazio retrorettale. Le cisti presacrali sono rare negli adulti. La maggior parte delle lesioni sono benigne. Le cisti possono essere uniloculari o multiloculari. Sono spesso asintomatiche. Questi tumori rappresentano un interesse diagnostico e terapeutico. Le cisti presacrali epidermoidi sono classificate come un tipo di cisti di sviluppo. Riportiamo il caso di una donna di 57 anni che si è presentata con una massa indolore, posteriore all' ano. Studi di imaging hanno mostrato una cisti multiloculare (65 × 100 × 60 mm) nello spazio retrorettale. La paziente è stata sottoposta ad un intervento chirurgico attraverso un approccio posteriore. Lo studio istologico ha rivelato una cisti epidermoide. Viene eseguita una revisione della letteratura di questo raro tumore.

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