Image findings of a tailgut cyst. Case report and short review of the literature

V.T. Skiadas, *M.D.*; **V. Koutoulidis,** *M.D.*; **H. Primetis,** *M.D.*; **Ch. Ladopoulos,** *M.D.*; **L.A. Moulopoulos,** *Assist. Prof.*; **A. Gouliamos,** *Prof.*; **L. Vlahos,** *Prof.*

Radiologic Department of Areteion Hospital, University of Athens, Athens (Greece)

Summary

We present a case of retrorectal hamartoma (tailgut cyst). Imaging findings on ultrasound, computed tomography and magnetic resonance imaging, pathologic findings, as well as the diagnostic pitfalls during the patient's management are documented. As it is a rare lesion with a non specific clinical presentation, it is usually misdiagnosed. Our aim is to present image characteristics of these lesions in all modalities and include retrorectal hamartomas in our differential diagnosis in patients with lesions with similar image findings.

Key words: Retrorectal hamartoma; Tailgut cyst; Ultrasound; Computed tomography; Magnetic resonance imaging.

Introduction

Developmental cysts are the most common congenital lesions encountered in the presacral region in adults. They include neurenteric cysts, epidermoid cysts, dermoid cysts and enteric cysts [retrorectal cystic hamartomas or tailgut cysts (TGC) and rectal duplication cysts] [1-3]. Retrorectal cystic hamartomas are uncommon retrorectal lesions that may appear in neonates or adults [4]. The rarity and the nonspesific clinical presentation of TGCs often lead patients to the operating room, due to misdiagnosis. We report a case of TGC and describe the ultrasound (US), computed tomography (CT), magnetic resonance imaging (MRI) and pathologic findings.

Case report

A 48-year-old woman was referred to our hospital due to dysuria and pain in the hypogastrium for the previous seven days. Transrectal examination revealed a palpable mass in the pelvis. Blood tests and urine examination were normal. Biochemical analysis for carcinoembryonic antigens revealed a high level of CEA (60.0 ng/ml - normal values: 0-3 ng/ml). Colonoscopy revealed a smooth stenosis of the distal 5 cm of the rectum with normal appearance of the rectal mucosa.

Transabdominal ultrasound examination of the pelvis revealed a 9 x 9.5 x 11 cm cystic mass in the presacral space with an obvious increase through transmission of sound. The mass displaced the bladder and uterus anteriorly. It appeared well defined and homogeneous in echotexture, except for a solid component with a small focus of calcification, adherent to the anterior wall of the lesion (Figure 1a). Several foci of calcification at the anterior cystic wall produced acoustic shadowing (Figure 1b). Color and power Doppler ultrasound revealed no vascularity of the wall. The primary diagnosis was that of an ovarian mass. The patient refused transvaginal sonographic examination.

CT of the abdomen and pelvis showed a well defined, homogeneous, large (10 x 10 x 11 cm) cystic mass in the retrorectal space, which anteriorly displaced the rectum, uterus and urinary bladder. Several foci of calcification were revealed at the periphery of the lesion, confirming the ultrasound findings. The mass was slightly hypodense in relation to the muscles (density values: 27-32 HU) and it extended into the second sacral foramina, which were widened (Figure 2a, b). Fat planes were preserved and there was no evidence of invasion of adjacent anatomical structures. Based on the CT findings, and in particular the association with the sacral foramina, the lesion was considered to be primarily a tumor of neurogenic origin.

MRI revealed a well-defined cystic mass with a low intensity capsule on all sequences, a fluid-fluid level and two mural nodules in the anterior aspect of the mass (Figure 3a-3d). Insinuation of the lesion in the second LT sacral foramen was confirmed (Figure 3c). A fat suppression technique excluded the presence of fat in the tumor. Contrast enhanced T1-weighted images showed mild enhancement of the nodules (Figure 3b). The lesion was considered to be primarily a retrorectal hamartoma. Abdominoperineal resection of the mass, the uterus and both ovaries was performed. No evidence if invasion of contiguous structures was confirmed at surgery.

Gross pathologic evaluation of the mass showed that the lesion was encapsulated. It consisted of multiple small cysts with hemorrhagic mucoid material. Two mural nodules were present. At microscopic examination the wall of the cyst and the mural solid components of the lesion were composed of fibrous tissue and smooth muscle fibers. Areas of calcification were identified at the wall of the lesion. The cyst lining was flattened and consisted of cuboid or cylindrical epithelial cells, with areas of squamous metaplasia. Xanthogranulomatous inflammatory infiltration was identified in many areas of the lesion. In one section of the specimen, a small number of striated muscle cells were found. No sites of malignant transformation were identified in the lesion.

Discussion

Tailgut cysts or retrorectal cystic hamartomas are rare, developmental lesions that were described for the first time by Middledorpf in 1885 [5]. However the term

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Fig. 1a

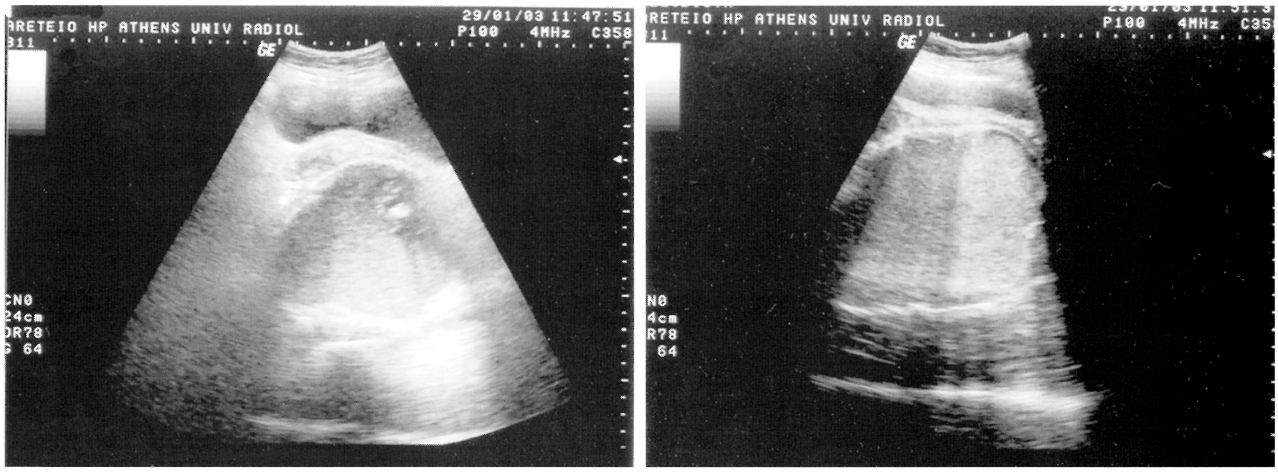


Fig.

Fig. 2a

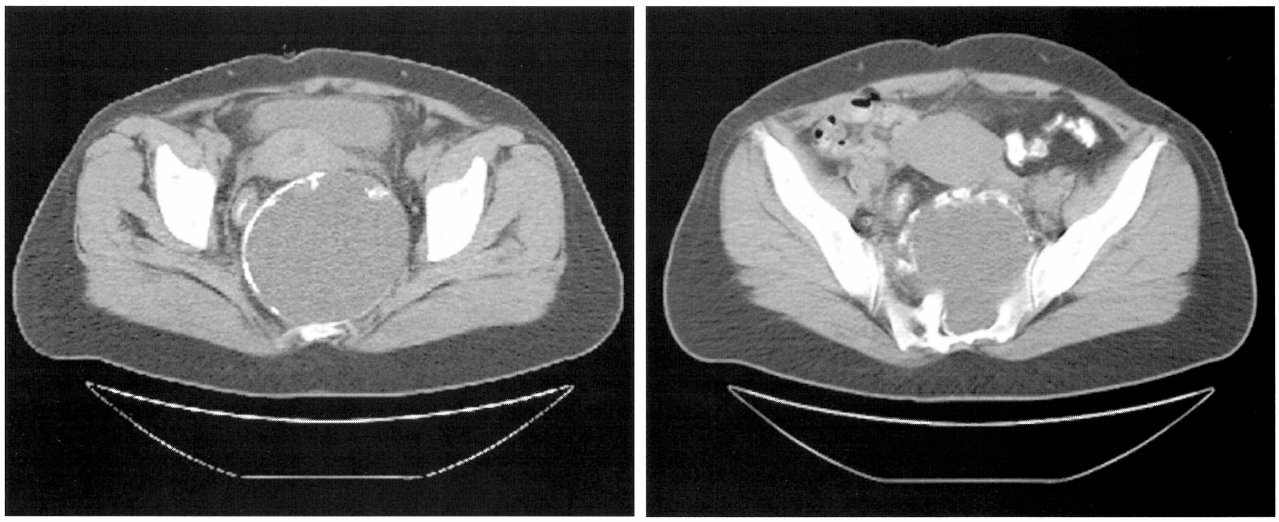


Fig.

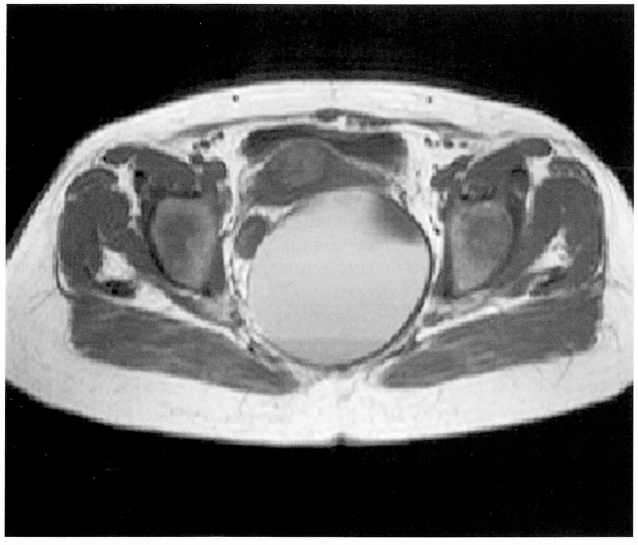
Figure 1. — (a) Axial pelvic ultrasound shows a well-defined cystic mass in the presacral space increasing through transmission of sound. The mass displaces the bladder and uterus anteriorly. A solid component with a small focus of calcification is identified. (b) Several foci of calcification at the anterior cystic wall produce acoustic shadowing.
Figure 2. — (a) Pelvic CT shows a well defined hypodense (27-32 HU) cystic mass suggestive of a cyst. Protrusion of the lesion into the second LT sacral foramina. (b) Calcification of a mural nodule at the anterior aspect of the mass and small foci of calcifications at the periphery of the lesion. Notice the displacement of the uterus, urinary bladder and rectum.

retrorectal cyst-hamartoma was introduced by Edwards much later in 1961 [6]. TGCs arise from remnants of the postanal primitive gut (tailgut), the most caudal part of the hindgut, which is normally expected to involute by the eighth embryonic week (35 mm stage - 56 days' gestational age) [7]. They are confined to the retrorectal space, between the rectum, sacrum and coccyx. They can be found at any age, from neonates to adults. Although they are congenital lesions, they become symptomatic when they attain a certain size. This is the reason why they are usually found in middle-aged patients, and they are more common in women. Hjermstad and Helwig in the largest series of 53 cases report a female to male ratio of 3.1:1 [1]. The reason for such a female predilection remains unclear.

TGCs are usually detected incidentally [8]. When they become symptomatic, patients may present with discomfort on sitting, constipation, lower abdominal or pelvic pain, rectal fullness, changes in caliber of stools, or urinary symptoms such as urinary frequency, dysuria or retention [7-10]. Patients may present with symptoms from superimposed complications such as infection with fistulization, rectal bleeding and malignant degeneration. Malignancy arising in these lesions is very uncommon. To the best of our knowledge 21 cases have been described in the literature. These few reported cases include 13 cases of adenocarcinoma, seven cases of carcinoid tumor and one case of sarcoma [1, 8, 10-18]. Retrorectal hamartomas are often misdiagnosed due to their atypical clinical features. A history of repeated surgical procedures before the correct diagnosis is not uncommon.

Sonography – transabdominal or transrectal – reveals a well-defined cystic mass behind the rectum, with or without increase via sound transmission, often with internal septations but with no evidence of invasion of adjacent structures. Low-level internal echoes can be identified [4, 19]. CT examination shows a discrete retrorectal

g. 3a



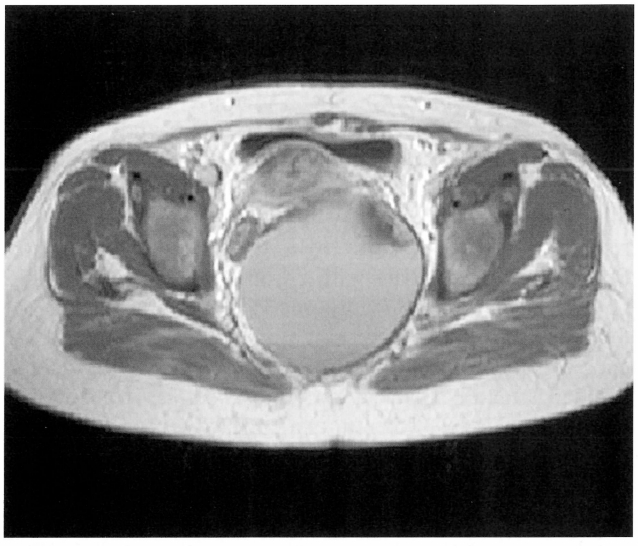
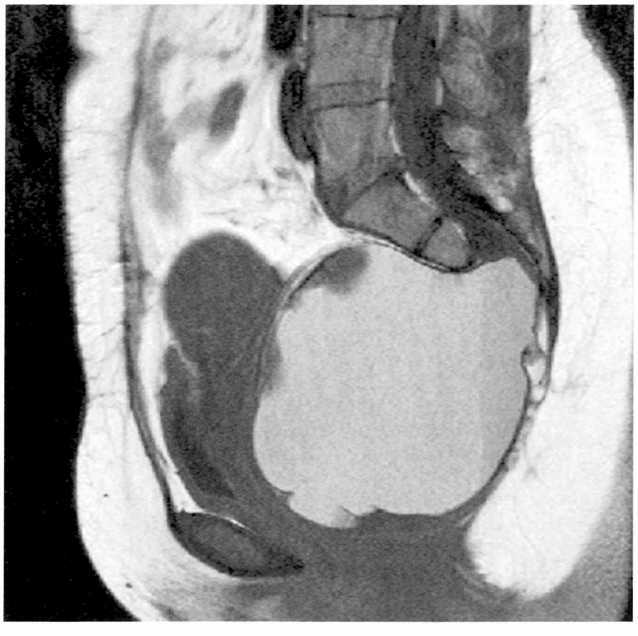


Fig. 3b

g. 3c



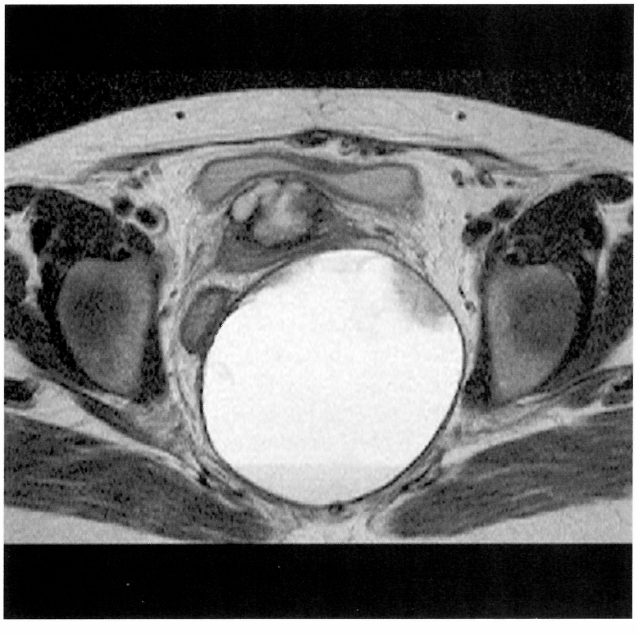


Fig. 3d

Figure 3. — (a) Axial T1-weighted (TR/TE 600/15), (b) axial T1-weighted (TR/TE 600/15) post GD, (c) sagital T1-weighted (TR/TE 600/15) and (d) axial T2-weighted (TR/TE 2500/80). MR images of the pelvis show a hyper-dense cystic mass with low intensity capsule, fluid-fluid level and mural nodules. Mural nodules show mild enhancement in image b.

mass with CT attenuation similar to that of water or soft-tissue. Keratinous, hemorrhagic or inflammatory content may result in higher CT attenuation values. No peripheral invasion is noted, unless there is concurrent infection or ma1ignant degeneration. In such case CT may show loss of discrete margins and fat planes around the lesion with or without involvement of contiguous structures [4, 8]. On MRI a tailgut cyst has a low signal intensity on T1- and high signal intensity on T2-weighted sequences. Internal septations are easily recognized when present. Fat-suppression techniques may exclude the presence of fat in the lesion. In cases with high protein content or internal hemorrhage the cysts will show high signals in T1-weighted sequences. Fluid-fluid levels may be observed as in our case. A thick fibrous wall secondary to repeated inflammations or surgical resections will show low signals in all MR sequences.

Macroscopically the lesions can be multi- or unilocular. They are usually confined to the presacral space and rarely involve the rectal wall. No communication with the rectal lumen is identified unless recurrent inflammations (resulting in fistulization) or previous surgical operations have taken place [1, 20]. Its maximum diameter ranges from 1 cm to 18 cm [7, 15]. TGCs are usually filled with fluid, which varies from a clear thin colorless liquid to a thick gelatinous substance, depending sometimes on the physical history of the lesion [1, 15]. Mural nodules can be identified in the lesion [10, 11, 15].

Microscopically a variety of epithelial types, found in the adult or fetal gastrointestinal track, may line the cysts. A single layer of epithelium without crypts to a wide range of epithelial types can be identified. Prasad *et al.* reported the presence of pancreatic cysts and islets of Langerhans in two of their cases. The most common type though, is that of squamous epithelium. In some cases this type of epithelium may represent metaplasia due to recurrent inflammation. Inflammation is a common finding in TGCs. Acute with chronic inflammation may be present and cause symptoms [7]. Xanthogranulomatous reactions may be present, almost always in the presence of squamous epithelium. A fibrous wall with disorganized smooth muscle fibers and absence of a double layer of muscles, submucosal and myenteric neural plexus is the typical finding. A thick fibrous wall can sometimes be present due to inflammatory changes [1, 8, 11, 21].

The differential diagnosis of a retrorectal lesion includes simple retrorectal epithelial cysts, dermoid and epidermoid cysts, teratomas, rectal duplication cysts, lesions of neurogenic origin, chordoma, mucinous adenocarcinoma, necrotic rectal leiomyosarcoma, cystic lymphangioma and pyogenic abscess [8-10]. Based on clinical and imaging characteristics of each lesion it is usually possible to establish the correct diagnosis.

The treatment of choice for TGCs is complete surgical excision [1, 7, 10, 11, 15]. The possibility of infection, recurrence of the mass, perianal fistulas and malignant transformation of any residual TGC remnants emphasize the importance of early and complete removal of the lesion. TGCs in neonates and young patients can be excised more easily and effectively than adult TGCs, as inflammatory changes lead to adhesion of the mass with adjacent anatomical structures [8, 10, 15]. A posterior approach is considered the best surgical method [22]. Removal of the coccyx and intraoperative decompression of the cyst allow better visualization and an easier surgical approach to the posterior elements of the cyst, especially in very large masses. Biopsy of the lesion is not indicated as it provides limited material and cannot lead to a definite diagnosis [7]. The prognosis of patients with TGC with or without malignant transformation is not well-defined due to the rarity of the lesion and the small number of reported cases [15].

Sonographic detection of a multicystic retrorectal mass with the features previously described, with absence of invasion of adjacent structures, should raise the suspicion of a tailgut cyst. Further imaging, either by CT or by MRI, is considered essential in order to establish the diagnosis. Detection of a solid component with contrast enhancement on CT or MRI should be considered suspicious for malignant degeneration of the cyst. Complete surgical removal is, in any case, mandatory.

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Address reprint requests to:
V.T. SKIADAS, M.D.
Makedonias 41
Ilion, 12321
Athens (Greece)