Retroperitoneal mass with ischiorectal fossa extension: diagnosis, clinical features and surgical approach. A literature review starting from a rare clinical case of primary retroperitoneal dermoid cyst

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Summary

Background: Primary retroperitoneal teratomas are rare and occur mostly in the sacrococcygeal area of children. They constitute less than 4% of all extragonadal teratomas with less than 120 cases having been reported, and only partly described in the retroperitoneum of adults. We describe an unusual case of a paravescical teratoma with ischio-rectal extension and its successful surgical management. *Clinical case:* A 24-year-old female was referred to our hospital with a history of pelvic pain, pressure and evidence of a pelvic-perineal tumor. Serial work-up disclosed a mass in thè left paravesical fossa that bulged out through the levator-ani muscle, in the left ischial-rectal fossa, altering the symmetry of the gluteal/perineal region. At surgery a cystic tumor, consistent with a dermoid, was completely excised from thè left paravesical fossa by means of a laparotomic approach. Pathological examination revealed a mature cystic teratoma. The postoperative course was smooth and the patient was doing well at two-year follow-up. *Conclusion:* This is the second reported case of paravesical dermoid cyst with ischio-rectal extension through the levator-ani muscle. Retroperitoneal teratomas are rare and difficult to early diagnose because of non specific signs and symptoms and should be considered in the differential diagnosis of a pelvic mass in adults. Solid and cystic morphology, fat signal and areas of calcification are some of the helpful features in diagnosing this neoplasia. Once the diagnosis is made, surgical removal is indispensable because of the indeterminate course of the disease. Prognosis depends on the histologic nature of teratoma. Patients with complete resection of benign teratoma have an excellent prognosis. Malignant teratomas, either with germ cell elements or with somatic elements, have a poor outcome.

Key words: Dermoid cyst; Mature/immature cystic teratoma; Retroperitoneal neoplasms; Paravesical fossa tumors; Ischiorectal fossa.

Introduction

Dermoid cyst, also defined as benign cystic teratoma, is a tumor comprised of a variety of parenchymal cell types representative, usually, of more than a single germ layer. Both terms are used as synonymous, although some authors continue to make a distinction, referring to dermoid cysts when indicating well arranged tumors with well-differentiated ectodermal and mesodermal derivatives surrounding endodermal tissues, and to teratoma when considering disorganized solid structures with immature components. They derive from a totipotential germ cell that can produce virtually any adult tissue (mature teratoma) and fetal tissue (immature teratoma), including abortive organs, limbs, hair, bones, and teeth.

The majority of teratomas are present in the sacrococcygeal region of infants, within the ovaries of adolescent females and within the testes of young males, but they have been identified in midline or paraxial structures such as mediastinum, retroperitoneum, pineal gland (and other intracranial and intraspinal sites) [1]. Sites for dermoid cysts also include the skin of the face, scalp and neck, and the floor of the mouth as well (Table 1).

Revised manuscript accepted for publication August 26, 2010

Primary retroperitoneal teratomas are rare and occur mostly in the sacrococcygeal area of children [2] (presumably due to a concentration of rich pluripotent cells at the end of the coccyx). They constitute less than 4% of all extragonadal teratomas [3] with less than 120 cases having been reported, and only partly described in the retroperitoneum of adults [1-7]. We describe an unusual case of a paravescical teratoma with ischio-rectal extension and its successful surgical management.

Case Report

In March 2003, a 24-year-old nulliparous was sent to the Department of Obstetrics and Gynecology, University Medical Centre, Ljubljana, because of mild pelvic pain, pelvic pressure, hesitancy of micturition and constipation on and off, with a bulging lower gluteal region on the left side. Her anamnesis was unremarkable and symptoms had been going on for ten months. On physical examination, a mass 10 x 10 cm in size filled the left side of the small pelvis; the tumor was slightly mobile, had well defined margins and bulged out in the left ischio-rectal fossa, making the appearance of the perineal and gluteal regions asymmetric.

The value of serum tumoral markers (AFP, p-hCG, CEA, CA 19.9, CA 125) were within the normal range as well other hematological and biochemical investigations. Sonography, either transabdominal either transperineal, showed an ipoechoic

Table 1. — *Anatomic location of dermoid cysts*.

	Anatomical sites
Gonadal	– *Ovaries* – *Testes*
Extragonadal	– *Skin* face, forehead, scalp, neck, midline region of trunk, penis,sacrococcygeal region – *Mucous membrane* floor of the mouth, tongue, hard palate, nasopharynx, vaginal wall – *Intracranial* pineal gland, eustachian tube, orbit, lateral ventricle – *Intraspinal* filum terminalis, conus medullaris or perispinal sites – *Mediastinum, chest* mediastinum anterior (mainly) and posterior, lung, thyroid, pericardium – *Abdominal viscera others than gonads* omentum, mesentery, bile ducts, stomach, large bowel, liver, pouch of Douglas, bladder, placenta, fallopian tube, uterus, round ligament of the uterus – *Retroperitoneal sites* kidney, diaphragm, iliac fossa, sacrococcygeal area (in adults mainly in the aortic retroperitoneum)

Table 2. — *Differential diagnosis of solid/cystic masses located in the small pelvis*.

	Anatomical sites
Extramedullary hematopoiesis	Sáenz-Santamaría J. Catalina-Fernandez I. [27]
Granuloma after inguinal herniorrhaphy	Azumi M. et al. [28]; Kise H. *et al*. [29]
Paravesical extension of psoas abscess	Oguchi K. et al. [30]
Aggressive angiomyxoma	Kaur A. *et al*. [31
Leiomyosarcoma	La Fianza A. *et al*. [32]
Schwannoma of the obturator nerve	Aubert J. *et al*. [33]
Lipoma	Hull W.B. *et al*. [34]
Liposarcoma	Fernicola A.R. *et al*. [35]
Lymphangioma	Irvine A.D. *et al*. [36]
Ectopic adrenal gland tissue	Rodríguez-Rivera García J. *et al*. [37]
Hemangiopericytoma	Sibert L. *et al*. [38] Dia A. *et al*. [39]
Angiofollicular hyperplasia of the lymph nodes (Castleman tumor)	Lehmann H.D., Seel R. [40]
Myelolipoma	Di Bonito L. *et al*. [41]

cystic mass (with anechoic portions) in the left pelvic-ischiorectal region with finely textured echoes within the fluid, few echogenic foci with posterior acoustic shadowing, and clear margins. No blood flow could be detected within the tumor using Doppler ultrasound; the uterus and adnexa showed no abnormalities and no ascitic fluid was detected.

Magnetic resonance imaging demonstrated an oval formation with fluid, soft tissue and lipid densities, and with clear margins filling the left ischio-rectal fossa (Figure 1). The mass displaced the uterus to the right and posteriorly, compressed the ab-estrinseco rectum and anus, and came in contact laterally with the internal obturator muscle and posteriorly with the left gluteal muscle. The tumor measured 10 cm cranio-caudally extending from the small pelvis to the subcutaneous fatty tissue of the left gluteal region. The relations between the mass and left levator-ani muscle were not clearly defined, the latter being crossed by the tumor or incorporated in it. Diagnostic laparoscopy was performed to evaluate an abdominal or transperineal approach. Laparoscopy disclosed a tumor mass filling the left paravesical fossa beneath the peritoneum of the large ligament. The mass became bigger, lifting the peritoneum and covering the paravesical space when pressure from the bottom upwards was applied to the gluteal region. The uterus, ovaries and tubes were grossly normal. An abdominal approach via Pfannenstiel laparotomy was performed, the left round ligament sectioned, and the left paravesical fossa developed; a well capsulated cystic tumor 12 x 10 cm in size filled thè left fossa extending deeply toward the pelvic floor. The cyst was tense, red and yellow in color, the surface was smooth, and some adhesions to the nearby organs were found (care was taken to avoid damage to the roots of the hypogastric vein and the obturator bundle). During dissection cyst wall rupture occurred and the sebaceous content mixed with hairs was carefully sucked from the lumen. The cystic wall was than grasped with a ring forceps allowing progressive traction on the mass. The tumor was excised completely and the pelvic floor integrity evaluated; the left levator-ani muscle was pushed downword (assuming the shape of a reverse dome) and the fibres were split allowing the mass to reach the adipose tissue of the ischiorectal fossa. On excision, the unilocular mass was filled with greasy material and tufts of hair, and the solid components were composed of fat, skin and bone fragments. A drainage tube was left in the remaining cavity for two days. Histologic diagnosis was a cystic teratoma filled with hair and sebaceous material and lined by a thin dermis-like wall containing sebaceous glands and others skin adnexa; no immature elements were present. The postoperative course was uneventful and the patient was well at two-year follow-up.

Discussion

Mature teratoma presents as a cyst lined by an epidermis-like epithelium with abundant sebaceous and sweat glands and dermal appendages, and contains variable mesoderrnal (bone, cartilage, smooth muscle, fibrous tissue), ectodermal (neural tissue, glia, retina, choroids, ganglia) and endodermal (gastrointestinal, bronchial, thyroid and salivary gland tissue) derivatives.

The exact histogenetic origin of teratomas is still controversial.

In the embryo, typically by the end of the third gestational week, uncommitted germ cells are normally sequestered in the hindgut region of the yolk sac near the allantois. These germ cells, in the fourth and fifth week of gestation, migrate by ameboid movement into the embryo along the dorsal mesentery of the yolk stalk reaching the urogenital ridge; during the sixth week, the germ cells develop into the spermatogonia or oogonia of the gonads. Some of these uncommitted cells could fail to reach the gonadal ridge, become misplaced, or migrate aberrantly; this migratory capacity may account for the multiple anatomic sites seen with these tumors [8]. These ectopic

Figure 1. — MRI: oval formation with fluid, soft tissue and lipid densities, and with clear margins filling the left ischio-rectal fossa. 1st) coronal view, T1 weighted; 2nd) coronal view, T2 weighted; 3rd) axial view. In MRI the mass can be clearly seen [1]; the alteration of pelvic-perineal anatomy [2].

germ cells retain their pluripotential capabilities and may develop into teratomas or any other type of germ cell neoplasia.

Gonadal teratomas appear to arise from reproductive cells that are the haploid descendants of embryonic germ cells, through some kind of parthenogenesis [9]. This theory (germ cell theory) is reinforced by chromosomal analysis demonstrating that the gonadal teratomas, although diploid, contain pairs of identical chromosomes rather than the normal pairs of maternal and paternal.

On the contrary, Wagner *et al*. [10] demonstrated that extragonadal teratomas arise from cells with a diploid chromosome; from this ascertainment they suggested that these teratomas arise from either premeiotic germ cells or pluripotent ectopic embryonal or extraembryonal cells. Since every somatic cell contains the full genetic code, theoretically, without being a "germ" cell, it could produce any other type of cell (embryonic cell theory). Although this is one of the principles of cloning, it allows an alternate theory of teratoma formation from ordinary somatic cells rather than from pluripotential germ cells.

A macroscopic classification of teratoma distinguishes two variants: cystic masses that are composed of mature elements and solid neoplasms that are more likely to contain immature tissues (mainly immature neuroepithelium whose amount is graduated by the grading scheme by Norris *et al*. [11]). The teratomas are also classified into three histopathologic categories: mature, made of well differentiated structures and tissues, immature, containing areas of primitive mesoderm, ectoderm or endoderm elements, and malignant containing frankly malignant tissues of germ cell origin (germinoma, yolk sac carcinoma, choriocarcinoma, endodermal sinus tumor). To these, pathologists add the "teratoma with malignant transformation" that include teratomas containing malignant non-germ-cell elements (squamous carcinoma, adenocarcinoma, melanoma, sarcomas) presumably derived from somatic tissue within the teratoma. A detailed teratoma classification system has been introduced by Olsen and Gonzales-Crussi in 1982 [8]; it allows, on the basis of the nature of tissues, their proportions and their characteristics, a better correlation of histologic findings and prognosis. According to this classification, a teratoma could be considered as mature if it contains up to 10% of undifferentiated tissue.

Primary retroperitoneal teratomas are rare and represent 1-11% of all retroperitoneal tumors [1], ranking third behind neuroblastoma and nephroblastoma in the pediatric population. The incidence has two peaks, in the first six months of life and in early adulthood [12] and is from two to three times more frequent in female than in males [13, 14]. In adults their prevalent locations include thè lombo-aortic retroperitoneum (with a left suprarenal predominance [15]), whereas pelvic retroperitoneal locations (sacrococcygeal area) are more frequent in children. It has been reported that 2% of dermoids undergo malignant transformation, mainly squamous cell carcinoma; this percentage seems to be higher for teratoma located retroperitoneally. Some authors have suggested considering mature teratomas as premalignant lesions and to carefully analyze and detect small areas of malignant change which indicate a high risk of recurrence [16]. Malignant change of teratoma is higher in adults than in children [14], with an incidence of 25.8% and 6.8%, respectively [15, 17]. However, Augè *et al*. [18] found a similar malignant rate (23.5%) among 34 retroperitoneal teratoma discovered during the first postnatal month. Until 1995 ten of the 40 reported cases in adults were malignant [19]. Retroperitoneal teratomas are usually asymptomatìc. The most common presenting manifestation is a very large space-occupying lesion responsible for compression of neighboring organs with consequent pain, discomfort, abdominal distension, gastrointestinal and genitourinary symptoms. While benign teratomas are usually diagnosed as an incidental finding, malignant ones are usually symptomatic because of rapid growth and progression. Rarely infection or acute abdomen due to a traumatic rupture of retroperitoneal teratoma, has been described [20-22]. The workup for dermoid cysts is largely radiographic; X-rays, sonography, CT and MRI are useful for the differential diagnosis in cases of space-occupying lesions of the retroperitoneum.

Plain radiographs are useful for the preoperative diagnosis, demonstrating suggestive calcifications resembling teeth or bone in up to 74% of cases or the calcific rim of the cyst (calcifications cannot be considered a sign of

benignity since 12.5-25% of calcified tumors are malignant [17]). If calcifications are absent, an opacity or radiolucent mass causing displacement of adjacent structures could be detected.

Ultrasound may show a solid, cystic, or mixed solid and cystic mass (echocomplex appearance) occasionally with a fat-fluid level; sonographic features that suggest the specific diagnosis of dermoid cyst are the presence of echogenic spots with acoustic shadows (teeth or bone fragments) or dermoid plugs (rounded polypoid soft tissue masses projecting into the lumen made of tufts of hair or of tissue overgrowth from the inner surface of the cyst). However, sonography has limited sensitivity because of poorly identified fat and calcifications which both suggest the diagnosis of teratoma. CT scan gives more specific information and usually shows a well marginated, multilobulated mass with both cystic and solid cornponents with fluid, fat, soft tissue and bone densities; the presence of a fatty portion, which represents sebum, is virtually pathognomic of these tumors.

MRI has advantages over CT as its better tissue contrast enables delineation of the internal components of the tumor more accurately, as well as determination of the relations of the mass to adjacent organs [23]. MRI better delineates the five portions that usually compose a dermoid: adipose tissue, bone, hair, sebum, and loose edematous fibrofatty tissue with skin. Fat is suggested by high-intensity signals on Tl-weighted images.

Angiography is an adjunctive tool useful in detecting the blood supply and presence of hypervascularity suggesting malignancy. Sometimes retroperitoneal teratomas may show abnormally high AFP [24], CEA [25], CA 19-9 [26] and CA 125 levels. Elevated serum markers are seen more often in patients with malignancy than in patients with immature or benign lesions. Postoperative rise in serum marker levels is a good indicator of recurrence only when they were also elevated preoperatively. Tumor masses of pelvic retroperitoneum should include every type of tumor arising from pelvic organs and developing extraperitoneally or from extraperitoneal structures, enlarged lymph nodes as well as non-neoplastic lesions including lymphoceles, abscess and hematoma. The differential diagnosis of cystic/solid tumors filling the lateral pelvic space should consider extramedullary hematopoiesis [27], granuloma after inguinal herniorrhaphy [28, 29], paravesical extension of psoas abscess [30], aggressive angiomyxoma [31], leiomyosarcoma [32], schwannoma of the obturator nerve [33], lipoma [34], liposarcoma [35], lymphangioma [36], ectopic adrenal gland tissue [37], hemangiopericytoma [38, 39], angiofollicular hyperplasia of the lymph nodes (Castleman tumor) [40], and myelolipoma [41]. It should be emphasized that teratoma is a surgical disease, with chemotherapy and radiotherapy having a relatively small role. The treatment of choice for retroperitoneal tumors is complete surgical excision, since definitive diagnosis is only achieved following histologic evaluation of the specimen. Other reasons are worsening symptoms and life threatening complications associated with the increasing mass effect as the tumor continues to grow, acute complications related to cystic rupture, or infection and malignant changes. Squamous carcinoma or malignant melanoma may develop from the skin components though this is rare; adenocarcinoma [42] and carcinoid [43] have recently been reported. The prognosis is excellent if complete surgical resection can be accomplished. When possible, the laparoscopie route potentially allows for a rapid recovery and minimal morbidity but requires laparoscopie skills and thorough preoperative planning.

Prognosis depends on the histologic nature of the teratoma. Patients with complete resection of benign teratomas have an excellent prognosis. Malignant teratomas, either with germ cell elements or somatic elements, have a poor outcome.

Conclusion

This is the second reported case [44] of a paravesical dermoid cyst with ischio-rectal extension through the levator-ani muscle. Retroperitoneal teratomas are rare and difficult to diagnose early because of non specific signs and symptoms, and should be considered in the differential diagnosis of a pelvic mass in adults. Solid and cystic morphology, fat signal and areas of calcification are some of the helpful features in the diagnosis of this neoplasia. Once the diagnosis is made, surgical removal is indispensable because of the indeterminate course of the disease.

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