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Case Report

Retrorectal cyst hamartoma (tailgut cyst) with malignant transformation

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Abstract

Background. Tailgut cyst [retrorectal hamartomas] is an unusual cystic lesion, located in the retrorectal space. It develops from post anal foetal gut remnants, present anterior to the sacrum and posterior to the rectum. A high index of clinical suspicion is required to diagnose this rare tumour. This malformative lesion should be distinguished from teratomas, mullerian cysts, anal gland cysts and duplication cysts of the rectum. The treatment of choice is complete surgical excision.

Case. A tailgut cyst is hereby reported, which had undergone malignant transformation, with development of an adenocarcinoma. Only 16 additional cases of tailgut cysts with adenocarcinoma were recovered, in an extensive search of world literature.

Conclusion. The identification of this rare entity provides an opportunity to examine this pathological rarity in the context of the few previously reported cases of this type.

Keywords: Tailgut cyst; Retrorectal cystic hamartomas; Adenocarcinoma

Introduction

Tailgut cyst (TGC) is a rare benign cystic lesion located in the retrorectal or presacral space. The rectum, defines this space anteriorly posteriorly, the sacrum, superiorly the peritoneal reflection, inferiorly the levator ani and coccygeus muscle, laterally the ureter and iliac vessels. In a study of 53 cases by Hjermstad and Helwig, a prior histological diagnosis was made in only two cases [1].

The incidence of malignancy in TGC is extremely rare, with most of them being either Adenocarcinomas or neuroendocrine tumours of varying grades. A search of the published literature yielded only 16 cases of Adenocarcinoma developing in tailgut cysts [2–13].

Embryology

During the 3.5 mm to 8 mm gestational stage (28 days gestational age to 35 days gestational age) of human development, the embryo possesses a true tail, which is caudal to the site of subsequent formation of the anus. The primitive hindgut extends into this tail. This caudal extension is called the tailgut or post anal gut (Fig. 1) and it usually completely regresses by 8th week of gestational age. The persistence of this as an embryological remnant is the probable origin of the TGC [1,10].

Although TGC may be clinically present in all age groups from neonates to adults, the anomaly is more commonly found in middle aged women [1,11]. The clinical presentation is frequently non specific and related to a mass effect pressing on surrounding tissues causing discomfort while sitting [12]. If the cyst becomes infected, it is often misdiagnosed as pilonidal cyst, anorectal fistula or recurrent retrorectal abscess [1,11–13].

Case report

A 57-year-old lady presented with complaints of low backache of 6 months' duration, which increased in severity in the sitting position.

On clinical examination a mass was palpable per abdomen. The mass was also palpable on per rectal and per vaginal examination. X-ray of the lower spine was unremarkable.

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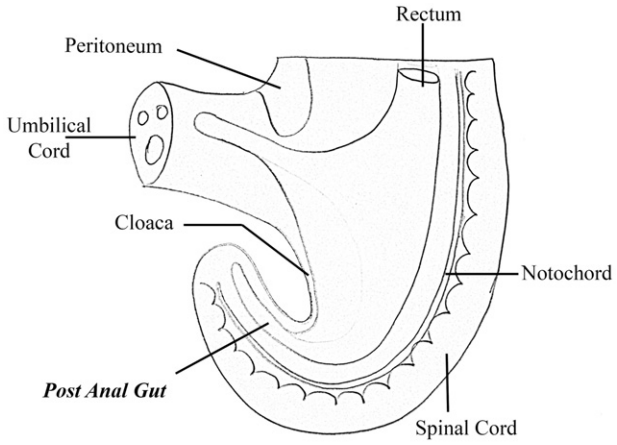

Fig. 1. Anatomical representation of the post anal gut during embryogenesis.

Ultrasonography was suggestive of a cystic ovarian tumour. Serum levels of CA-125 were normal. At laparotomy the uterus, fallopian tubes, and both the ovaries were found to be normal. The mass was present in the retroperitoneum in the retro rectal/presacral space. It was densely adherent to the sacrum and coccyx and was also adherent to the perirectal fat. As the cyst was large it was aspirated to decompress it and then was carefully dissected and removed intact.

Pathology

The smooth surfaced mass measured 12×10×8 cm and weighed 370 g. Cut surface showed a thick walled cyst divided into multiple locules by septae (Fig. 2) and containing thick turbid greenish brown fluid and slough. The inner surface showed a few papillary excrescence and the walls were thick and irregular varying in thickness from 0.4 to 1.0 cm.

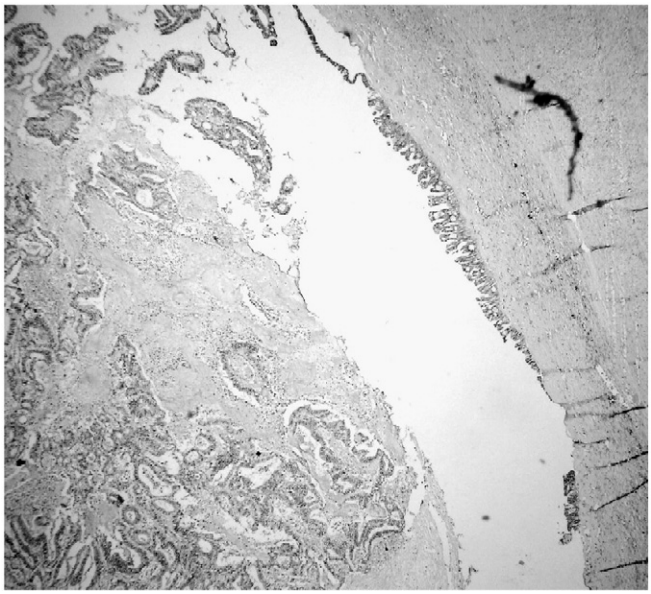

Fig. 2. Adenocarcinoma in the tailgut cyst, H&E stain, 100×.

On microscopy the cyst and its septae were lined by epithelium, which varied from being stratified squamous in places, transitional (cloacal) and columnar, mucinous in others (Fig. 2). Large areas were ulcerated and replaced by siderophages, and chronic inflammatory cells.

The mucinous epithelium showed gradual dysplastic and carcinomatous transformation. It developed papillary and cribriform structures lined by columnar to cuboidal cells with moderately pleomorphic mitotically active nuclei. Variably sized invasive malignant glands with abundant mucin formation formed which then invaded almost through the full thickness of the cyst wall (Fig. 2).

The fibrous wall of the cyst and its septae contained fascicles of smooth muscle. Areas of necrosis, haemorrhage and calcification were seen in the wall.

A diagnosis of a grade 2 adenocarcinoma without full thickness invasion in a tailgut cyst was offered. The cyst had been removed intact.

The postoperative period was uneventful. CEA done in the postoperative was found to be within normal range. CA 19-9 was normal.

MRI abdomen and pelvis (post surgery) was unremarkable. The number of such cases is too few for an established chemotherapy protocol. Therefore the regimen for rectal carcinomas was offered to her (6 cycles of chemotherapy comprising of 5-fluorouracil and leucovorin). However she refused chemotherapy, and at her last follow up 10 months after surgery was found to have multiple metastasis in the liver.

Discussion

The tailgut cyst or retrorectal cyst hamartoma is an unusual malformative cystic lesion, which develops in the retrorectal space. This is a potential space, which lies between the rectum

Table 1
Cystic lesions in the pre-sacral region

Type of cyst	Lining of the cyst	Composition of the cyst wall	Other pertinent features
Tailgut cyst	Variety of cloacal epithelia	Smooth muscle	Anatomical location
Epidermal cyst/ dermal cyst	Only squamous epithelium with keratin/dermal appendages	No smooth muscle	
Teratoma	Presence of dermal appendages	Variety of elements with organoid organization	Usually seen in pediatric population
Rectal duplication cyst		Well-developed muscle layers with nerve plexus in between	Develops from a later stage of embryogenesis
Mullerian cyst	No transitional epithelium	Mesenchyme can be induced to lay down smooth muscle in wall	Other foci of endometriosis in pelvis
Anal gland cyst			Associated with anal sphincter rather than retrorectal space

and the sacrum and is bounded superiorly by the peritoneal reflection and inferiorly by the muscles of the pelvic floor.

The tailgut is a post cloacal extension of the embryonic gut (Fig. 1) and in the normal course of embryogenesis involutes completely along with the embryological tail, by the 8th week of gestation. The tailgut cyst is believed to derive from a persistence of embryologic remnants of this post anal gut (tailgut).

Though this malformative lesion can develop in any age group, it is most commonly reported in women in the 4th to 6th decade. They present most often to the gynecologist with complaints of backache or vague obstructive symptoms related to the effects of a mass in the pelvis. These cysts are also often incidentally detected during imaging for unrelated causes.

The cysts that develop from these tailgut remnants at the time of detection range in size from 2 cm to 12 cm and have a fairly standard morphology. They are multiloculated cysts filled with debris and mucin and lined by a variety of cloacal epithelia ranging from stratified squamous to cuboidal, transitional, seromucinous and intestinal. Ciliated epithelium is also sometimes seen and as it is known to occur in the embryonic gut is not exclusively indicative of respiratory origin. Poorly developed fascicles of smooth muscle are invariably present in the wall.

Other cystic lesions that also develop in this region and to be considered in the differential diagnosis are teratomas, mullerian cysts, endometriotic cysts, epidermal cysts, rectal duplication cysts and anal gland cysts. The pertinent features to differentiate these cysts are enumerated in Table 1.

The tailgut cyst predominantly benign can become secondarily infected and is then mistaken for a retro rectal abscess or perineal abscess, for which repeated attempts at drainage are sometimes carried out.

Malignant transformation is an unusual complication. Most of the neoplasms that develop are adenocarcinoma of the garden variety, though neuroendocrine tumors of varying grades have also been reported.

In these adenocarcinomas a dysplasia–carcinoma sequence is often noted in the cyst lining. Like other colorectal carcinomas, these tumours are often positive for CEA [2,11,13] and serum CEA levels can then be used to monitor their progression. Positivity for p53 has also been noted in both the dysplastic and malignant epithelium [9,12].

Prognosis for tailgut cysts with malignant transformation depends on the status of complete surgical resection and tumour histology and grade. Local recurrences and distant metastasis have been occasionally reported.

In conclusion, tailgut cysts are rare malformative lesions, which probably remain asymptomatic. However they may grow to considerable size and present in adult life with obstructive symptoms, secondary infection and rarely with malignant transformation.

A high index of suspicion is required for the diagnosis of these cysts. The anatomic location and the variety of cloacal epithelia that form the lining, support an origin from the tailgut. Complete surgical excision at the time of diagnosis is the treatment of choice, as malignant transformation cannot be pre-operatively predicted.

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