A tailgut cyst in a patient with myelomeningocele – An association or a coincidence? – A case report.

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Case Report

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Abstract

Tailgut cysts (TGC) are rare lesions occurring in the retrorectal space, a potential space anterior to the sacrum and coccyx and posterior to the rectum. They are developmental lesions that have potential for malignant transformation into neuroendocrine tumors, carcinomas, or adenocarcinomas. However, they present with non-specific complaints and are often challenging to diagnose and can lead to misdiagnosis and delay of treatment. We present a case of a 27-year-old female patient with history of a myelomeningocele who was found to have a left sided pre-sacral cystic mass. Histologic examination of the lesion following surgical removal were consistent with TGC. The presence of a TGC in a patient with myelomeningocele is a rare occurrence. There is some evidence that disturbances in the Sonic Hedge Hog (SHH) signaling pathway can lead to abnormal branching and overgrowth of the notochord contributing to abnormal separation from the hindgut endoderm. Clinicians should have a heightened clinical suspicion for a TGC in patients with known neural tube defects who present with a perirectal mass lesion.

Introduction

Tailgut cysts (TGCs) or retrorectal cystic hamartomas are rare lesions in the retrorectal space, a potential space anterior to the sacrum and coccyx and posterior to the rectum [2]. The rarity and frequently nonspecific complaints of these tumors can lead to misdiagnosis.

Case Report

A 27-year-old female was found to have a left sided pre-sacral cystic mass on imaging following surgery for an ovarian mass. The patient endorsed low back pain, lower extremity weakness and leg pain in a non-dermatomal distribution. Additional past surgical and medical history included myelomeningocele repaired postnatally, hydrocephalus requiring a ventriculoperitoneal shunt, and a thoracic arachnoid cyst requiring fenestration surgery. On physical exam, she demonstrated diminished sensation to light touch on the left side, focal distal muscle weakness and brisk reflexes bilaterally.

On MRI, a multilobulated mass measuring about 4.0 x 4.5 cm was seen in the pre-coccygeal region. The mass was hypointense on T1- and intermediate intensity on T2-weighted images. No enhancement was seen with contrast administration (Fig. 1). Given that patient was symptomatic, it was decided to perform an excisional biopsy of this lesion to establish the diagnosis.

A midline sacral laminectomy using neuro-monitoring was performed. Following skin incision and soft tissue dissection, the anococcygeal ligament and levator ani were identified and subsequently divided. The coccyx was removed. Meticulous dissection created a plane between the lesion and an associated dermal sinus tract and the adjacent para-coccygeal muscles, ultimately excising the cystic lesion and associated dermal sinus tract en bloc.
The surgical pathology specimen consisted of a cystic mass measuring 6.0 x 4.4 x 4.2 cm$^3$. This was larger than was anticipated per imaging findings obtained eight months prior. On gross examination, the cyst wall was smooth with thick grey contents. Upon histologic examination, the lesion was multilobulated, lined predominantly by squamous epithelium and focally by stratified columnar epithelium with cilia (Fig. 2a). The cyst contents were composed of keratinous and cellular debris with neutrophils. The soft tissue surrounding the cyst showed areas of foreign body giant cell inflammatory reaction and cholesterol clefts. Glomus bodies, aggregates of bland cells with round nuclei and abundant eosinophilic cytoplasm, were noted within the cyst walls (Fig. 2b). These cells stained positively with smooth muscle actin. The location and histology were supportive of the diagnosis of a TGC.

**Discussion**

TGCs occur most commonly in the retrorectal space but may also occur in perirenal or perianal regions. During normal development, regression of the embryonic tail and hindgut occurs by the eighth week. When regression is incomplete, a vestigial remnant such as a TGC may persist. TGCs are more common in women with a 3:1 ratio to men and are asymptomatic in approximately half of the cases. Age at diagnosis tends to be in the adult patient, however it varies widely. Clinical symptoms are often nonspecific. Pain is the most commonly reported symptom, especially low back pain with or without radicular symptoms. Other symptoms include rectal or anal pain, constipation, swelling, and chronically draining wounds. Given the rarity of TGCs and the nonspecific nature of their presentation, diagnosis is quite challenging. This may lead to patients being misdiagnosed and undergoing multiple unnecessary procedures.

Radiographically, plain films may be able to demonstrate sacrococcygeal abnormalities such as bony defects. However, the imaging modality of choice for identification of retrorectal hamartomas is CT or MRI. On CT, these lesions appear well-marginated, thin-walled, either uni- or multi-locular, hypodense, and non-enhancing in the retrorectal space. They are rarely associated with thin calcifications. On MRI, TGCs appear as well circumscribed, thin-walled, hypointense lesions on T1 and hyperintense on T2. Septations may be visible on T2 weighted imaging [2; 10].

Histologically, TGCs demonstrate an epithelial lining composed of stratified squamous, columnar, ciliated columnar, mucinous gastric, cuboidal and/or transitional cell types. Often disorganized areas of smooth muscle tissue are found as well [7].

The risk of malignant transformation into neuroendocrine tumors, carcinomas, or adenocarcinomas necessitates complete surgical excision with negative margins as the treatment of choice [5]. Anterior, posterior or combined anterior-posterior approaches have been used [1]. More recently, trans-anorectal approaches have also been described [6]. Anterior approaches tend to be used for higher lesions and posterior approaches typically for lesions below the sacral promontory. Early diagnosis and resection may allow for a complete resection with lower risk of incontinence and overall morbidity [5].
The presence of a TGC in a patient with myelomeningocele is a rare occurrence. In a literature review, we found only two similar cases of lumbosacral vertebral anomalies in association with hindgut malformations. Kemp et al. reported a case of an anterior sacral meningocele seen in conjunction with a TGC, tethered spinal cord and syringomyelia [3]. Mitsuyama et al. described a TGC and thickened filum terminale giving rise to a tethered spinal cord [8].

Developmentally, the notochord is thought to play a pivotal role in both the cranial-caudal and ventral-dorsal organization of the adjacent embryologic tissue including the spinal cord, vertebral column, and hindgut. There is some evidence that disturbances in the Sonic Hedge Hog (SHH) signaling pathway can lead to abnormal branching and overgrowth of the notochord contributing to abnormal separation from the hindgut endoderm. In studies on ethylene thiourea treated fetal rats to induce anal rectal malformations, neural tube defects were present in 24 of 34 embryos [9]. Supporting this theory, a mouse model has been developed showing that anorectal malformations (including teratomas and enteric cysts) and anterior sacral myelomeningoceles share a common embryologic pathway that is dependent on the extent and duration of abnormal adhesion between the neural groove and future gut [4].

**Conclusion**

The presence of a TGC in a patient with myelomeningocele poses the question of whether there is a true association between them. There have been two case reports of lumbosacral vertebral anomalies presenting in association with hindgut malformations and experimental models that suggest both pathologies may share some similar developmental pathways.

The nonspecific symptoms that accompany TGC presentations may result in underdiagnosis. Clinicians should have a heightened clinical suspicion for a TGC in patients with known neural tube defects who present with a perirectal mass lesion.

**Declarations**

*Ethical Approval* – Consent for publication was obtained verbally from the subject of this case study. Written consent was not obtained as the case was determined to meet criteria for IRB exemption.

*Competing interests* – There are no financial or non-financial competing interests to declare.

*Authors’ contributions* – Timothy Beutler and Michelle De Witt conducted the literature review, wrote the main manuscript text, and prepared figure 1. Aylin Padir assisted in writing the main manuscript text and preparing the text for submission. Maria Caicedo Murillo facilitated the pathology review and prepared figure 2. Jiri Bem and Satish Krishnamurthy were lead surgeons on the case. All authors reviewed the manuscript.

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References


Figures
Figure 1

Pre- (1a) and post- (1b) contrast axial T1-weighted MRI images demonstrated a multi-lobulated hypointense mass in the pre-coccygeal region with no enhancement on contrast administration.

Figure 2
Histologic Examination with Hematoxylin and Eosin Stain (2a) Multiloculated cyst lined by stratified epithelium with large amount of keratin material within the lumen [black arrow] and several areas of the cyst lined by ciliated columnar epithelium [blue arrow] (2b) Stratified squamous epithelium cyst lining with keratin debris at higher magnification (2c) Ciliated columnar epithelium cyst lining at higher magnification [black arrow] (2d) Glomus body within the cyst wall comprised of a perivascular collection of bland cells with round nuclei and abundant eosinophilic cytoplasm which were immunohistochemically positive for smooth muscle actin