

# Adenocarcinoma in a retrorectal cystic hamartoma

## A case report and literature review

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### Summary

Retrorectal cystic hamartomas are rare congenital lesions derived from the vestigial portion of the embryonic hindgut. These lesions present in a vague manner and so mimic the presentation of more common mass lesions in the pelvic region. They most frequently afflict middle-aged women, causing vague pelvic pains, discomfort with defaecation or sitting, and altered bowel function. Malignant transformation within these lesions is extremely rare.

We describe a case of adenocarcinoma within a retrorectal cystic hamartoma, initially manifesting in a 54-year-old woman and recurring over a 5-year period. A computed tomography (CT) scan and pathology reports are included in the study, and findings at operations are discussed. We also include a comprehensive review of the literature.

### Case report

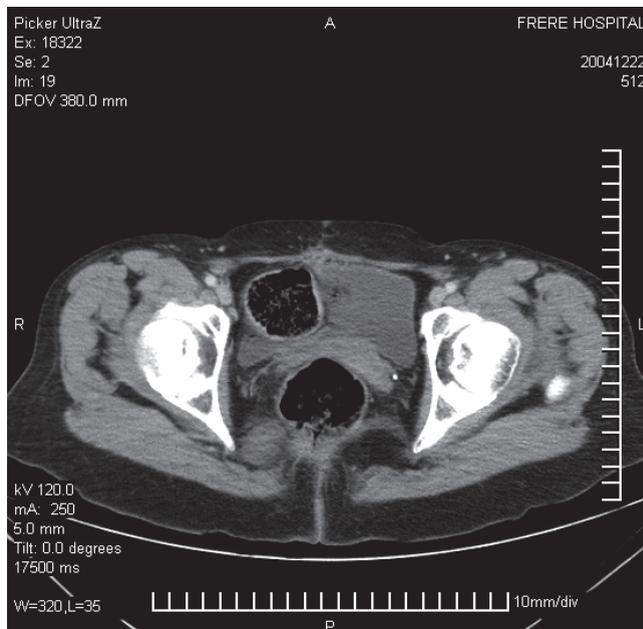
A 54-year-old woman was referred from her local clinic, complaining of a persistent, thick, yellow vaginal discharge, urinary difficulty and lower abdominal pain. Clinical examination confirmed that the cervix and rectum were displaced anteriorly by a tender, tensely cystic mass. An ultrasound scan showed a 12 cm well-circumscribed mass in the pouch of Douglas, containing small calcified areas. No abnormalities were discovered on routine haematological or biochemical investigations.

The patient underwent laparotomy for excision of the presacral mass. A 15 x 25 cm cyst was present in the retrorectal area, with a thin capsule and jelly-like mucinous content. There was a satisfactory plane around the cyst, which was finely tethered to sacrum and coccyx.

On microscopic examination, the loculated cyst was found to be lined by cuboidal columnar epithelium merging with areas of thin squamous epithelium, and in one area there was a well-established invasive adenocarcinoma. A pathologist at another unit stated that the histological appearance resembled that of ovarian metastases, but no investigations or tumour markers (CA125) supported this assertion. However, this did result in CA125 rather than carcinoembryonic antigen (CEA) being used for monitoring during the first post-operative year.

The patient completed six courses of chemotherapy (cisplatinum/cyclophosphamide), despite episodes of severe pancytopenia, an *E. coli* urinary tract infection and septicaemia.

More than a year later she was referred back to us. She had experienced 3 months of increasingly severe pelvic pain, especially when sitting. A tender mass could be felt anterior to the sacrum at the tip of the finger on rectal examination. A computed tomography (CT) scan (Fig. 1) showed a small soft-tissue mass, 3 x 4 cm in diameter, anterior to the sacrum at the level of S3 and associated with partial destruction of the sacrum, suggestive of tumour recurrence. There was no evidence of metastatic disease. The CEA value was elevated (51.5 ng/ml).



**Fig. 1.** CT scan showing posterior rectal wall thickening with effacement and sacral destruction. This extends along a fat plane adjacent to the rectum towards the right.

A transverse sacro-coccygeal incision was made and further access obtained by the removal of the coccyx and S5 vertebral body. A 4 x 5 cm cystic structure was found to be

invading the anterior surface of the sacrum. The content had a jelly-like consistency. The cystic structure was removed piecemeal owing to the bone invasion. Histological examination confirmed the recurrence of invasive adenocarcinoma, i.e. malignant recurrence within the tailgut cyst. The postoperative course was satisfactory, and the patient remained well for 2 years.

In early 2007 she presented with lower abdominal pain, constipation, significant weight loss and abdominal distension, and abdominal radiographs demonstrated complete large-bowel obstruction. At laparotomy extensive recurrence in the pelvis was found and a palliative sigmoid loop colostomy was performed.

## Discussion

A hamartoma is an excessive focal overgrowth of normal mature cells in an organ composed of identical cellular elements. The retrorectal cystic hamartoma, also referred to as a tailgut cyst, is believed to originate from the vestigial elements of the embryonic hindgut. When the embryo is between 4 and 8 mm in size, it possesses a true tail. This tail reaches a maximum diameter on day 35. By the time the orifice of the anus has developed cephalad to it (by day 56), the post-anal gut and neurentic canal have disappeared.<sup>1,2</sup>

Owing to the rarity of the condition and its nonspecific presentation, diagnosis is frequently late.<sup>1,3</sup> The largest series of retrorectal cystic hamartomas described is of 53 cases over 35 years (Armed Forces Institute of Pathology, Washington, DC).<sup>4</sup> A further 43 cases were described between 1988, the year this series was published, and 2005.<sup>5</sup>

These lesions are most common in middle-aged women.<sup>1,6,7</sup> The mass is frequently discovered during routine examinations, during childbirth, or in the course of investigation of other unrelated complaints. Other presentations include rectal, buttock or perianal discomfort, particularly while sitting, as well as rectal bleeding, pain with defaecation, lower back pain, constipation, urinary frequency or retention or even menstrual irregularities.<sup>1,6,8,9</sup> Rectal examination usually reveals a well-circumscribed structure compressing the rectum posteriorly.<sup>6,10</sup>

They may become secondarily infected, and can present like pilonidal cysts, anorectal fistulas or abscesses.<sup>1,11</sup> Malignant change within these lesions is extremely rare.<sup>1,6,11-15</sup> The vague nature of presentation may simulate that of many pelvic or lower abdominal mass lesions. It is important to exclude these lesions before sorting through the spectrum of congenital, inflammatory, neurogenic or osseous tumours that occur in the retrorectal space. Teratomas, epidermoid and dermoid cysts, rectal duplication cysts and anal cysts may share similar characteristics with a tailgut cyst.<sup>8,11,16,17</sup>

Important differential diagnoses are excluded pathologically by considering the structure and epithelial lining of the cysts in question. Epidermoid and dermoid cysts are usually unilocular structures, lined by stratified squamous epithelium. Dermoids have dermal appendages. Duplication cysts are also unilocular, but are lined by epithelium that simulates the normal gut mucosa. The main distinctive feature of these cysts is a well-formed continuous two-layered muscular wall with its own nerve plexus.<sup>1,3,4</sup>

Tailgut cysts, meanwhile, are frequently multicystic or multiloculated and may be lined by a wide variety of epi-

thelia, including stratified squamous, transitional, stratified columnar and gastric types. They may contain focal areas of smooth-muscle fibres, randomly positioned.<sup>1,6</sup> Most cases are located within the retrorectal (presacral) space.<sup>1</sup>

The most useful investigation is the CT scan,<sup>18</sup> which illustrates a well-defined retrorectal structure. The presence of local invasion implies malignancy. Calcification generally suggests other diagnoses, although it has occasionally been described in these lesions, especially with carcinoma in situ.<sup>1,3</sup> Magnetic resonance imaging shows a well-circumscribed lesion at T1 or T2 weighting, that is homogeneously hyperintense at T2 weighting or with a multilocular honeycomb appearance with thin internal septation.<sup>14,19</sup>

These lesions should be excised early to reduce the risk of malignant change and other complications.<sup>1,6,19</sup> Choice of route will depend on the local anatomy.<sup>7,20</sup> The lesions are often densely adherent to surrounding structures and require careful dissection.<sup>1,6</sup> They are usually between 2 and 10 cm in size, soft and well-circumscribed, and multicystic or multiloculated. Aspiration of larger lesions has been used to aid excision.<sup>1,20</sup>

The cysts are filled with fluid of variable density; the colour may range from yellow to green.<sup>1,3</sup> The lining epithelia vary considerably, although stratified squamous epithelium is most commonly found. There is a thin layer of fibrous tissue overlying the epithelium with occasional bundles of well-formed smooth-muscle fibres. This layer may also be densely infiltrated by inflammatory cells.<sup>1,6</sup>

At the time of writing malignant change within a retrorectal cystic hamartoma had been documented in only 31 cases. The majority were adenocarcinomas or neuro-endocrine tumours, although there have also been cases of carcinoid tumours described. In a recent review of the literature, Killingsworth calculates that there have been at least 17 cases since 1988.<sup>1,5,13,21</sup>

Adenocarcinoma usually originates within the cyst, and thereafter infiltrates the surrounding stroma. Well-formed glandular structures have been identified in specimens and p53, Ki-67 and p21 have been located by immunohistochemistry.<sup>5</sup> The dysplasia-carcinoma sequence is consequently assumed to occur within the lesion.

Other published case reports confirm the importance of early excision (not least because it can seldom be determined whether the lesions are benign or malignant pre-operatively), the presence of small nodules of cancer in otherwise benign cysts, and the value of CEA in follow-up.<sup>1,6,12,15,21</sup>

## Conclusion

Retrorectal cystic hamartomas are rare congenital lesions derived from the vestigial portion of the embryonic hindgut. These lesions present in a vague manner and so mimic the presentation of more common mass lesions in the pelvic region. They most frequently afflict middle-aged women, who present with vague pelvic pains, discomfort with defaecation or sitting, and altered bowel function. A presacral cystic mass is often palpable on routine rectal examination and further delineation is possible with CT scanning. Early careful surgical excision is required to prevent complications and malignant transformation. CEA levels should be monitored during follow-up of these lesions.

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