Retrorectal Cystic Hamartoma

Report of 5 Cases With Malignancy Arising in 2

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• ***Background.*—Retrorectal cystic hamartomas, or tailgut cysts, are rare congenital lesions that typically present as presacral masses. These lesions are frequently clinically unrecognized and misdiagnosed. Malignant change is extremely rare. Only 10 additional cases with associated malignancy were recovered from the literature. We describe the clinicopathologic features of 5 cases, including 2 cases with malignant transformation.**

***Results.*—All patients were women (age range, 36–69 years). The most common symptoms were pain with defecation and rectal bleeding. One patient was asymptomatic. All lesions presented as multicystic presacral masses and all were surgically resected. The lesions varied in size from approximately 2 to 12 cm (average, 9.5 cm) and overall had similar histology composed of a variety of epithelial linings (stratified squamous, transitional, and simple or ciliated pseudostratified columnar). Skin adnexa, neural elements, and heterologous mesenchymal tissue, discriminators between retrorectal cystic hamartoma and teratoma, were not identified. Arising in association with the cysts was a focus of adenocarcinoma in one case and a neuroendocrine carcinoma in another.**

***Conclusions.*—The clinical diagnoses in our cases were often delayed, which in part may be due to unfamiliarity with this entity. The main diagnostic difficulty is distinction from presacral mature cystic teratomas and rectal duplication cysts. Tailgut cysts require complete surgical excisions to prevent future recurrences and to preclude possible malignant transformation. Meticulous gross examination and adequate sampling are important to document the exact nature of these cysts and to rule out possible coexisting malignancies, which may be focal.**

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Retrorectal cystic hamartomas are uncommon lesions that may appear in the presacral (retrorectal) space in infants or adults. Most descriptions are anecdotal; the largest series of 53 cases was collected over a 35-year period by Hjermstad and Helwig[1] at the Armed Forces Institute of Pathology, Washington, DC. Only 2 of their cases had an initial pathologic diagnosis of retrorectal cystic hamartoma; most of the cases were initially diagnosed as a variety of congenital cysts. The incidence of malignancy in retrorectal cystic hamartomas is extremely rare. After extensive review of the world literature, we could identify only 10 other cases of malignancy arising in tailgut cysts. We hereby describe the clinicopathologic features of 5 cases of retrorectal cystic hamartomas, including 2 cases with malignancy. Cases with malignancy reported in the literature are reviewed, and the differential diagnosis of retrorectal cystic lesions is also discussed briefly.

MATERIALS AND METHODS

From 1981 to 1997, 5 cases fulfilling the criteria of retrorectal cystic hamartomas were identified from the surgical pathology and consultation files at Henry Ford Hospital, Detroit, Mich. One of these cases had a focus of adenocarcinoma, and a neuroendocrine carcinoma was found in another. We reviewed the hematoxylin-eosin– and immunoperoxidase-stained slides as well as the medical records of these 5 cases.

RESULTS

Clinical Data

Table 1 lists the clinical features of these 5 cases. All patients had barium enemas and sigmoidoscopies, which revealed no mucosal abnormalities except for mild diverticulosis in 2 cases. Digital rectal examination in all cases showed a nontender, extrinsic, well-defined presacral mass compressing the rectum. The lesions were often densely adherent to surrounding structures, requiring sharp surgical dissection. Occasional cysts were entered inadvertently during surgery, and some of the larger cysts required intraoperative aspiration to reduce their size. Due to the findings of a neuroendocrine carcinoma in case 5, a second surgery was performed to ensure complete removal of the tumor. There were occasional small cysts within dense fibrous tissue, but no residual neuroendocrine carcinoma.

Pathologic Features

The lesions were often removed in a piecemeal fashion because of dense fibrous adhesions to surrounding structures; consequently, their exact size could not be determined. Based on the dimensions of the largest intact cyst received, the masses varied from 2 to 12 cm. Grossly, all lesions were soft, well-circumscribed, multicystic or multiloculated masses. Externally, the cyst walls showed ad-

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Table 1. Summary of Clinical Features of the Five Cases

Case No.	Age, y/ Sex	Clinical Features	Radiology	Surgery and Follow-up
1	50/F	Rectal discomfort and pain for 2 mo	CT: large presacral mass extending cephalad	Surgical excision; ANED 3 y postoperatively
2	36/F	Asymptomatic retrorectal mass discovered during routine physical	MRI: 9.5 × 9.2 × 8.8-cm presacral multilocular cystic mass with a solid area	Surgical excision; ANED 2 y postoperatively
3	43/F	Painful bowel movements and low back pain for 12 mo	Radiograph: abdomen unremarkable except for ?erosion of sacrum	Surgical excision; ANED 17 y postoperatively
4	54/F	Rectal bleeding for 6 mo	MRI: 2-cm presacral cystic mass	Surgical excision; ANED 3 y postoperatively
5	69/F	Painful bowel movements and mild rectal bleeding for 12 mo	CT: 4-cm presacral cystic mass	Surgical excision; ANED 2 y postoperatively

* CT indicates computed tomographic scan; MRI, magnetic resonance imaging; and ANED, alive with no evidence of disease.

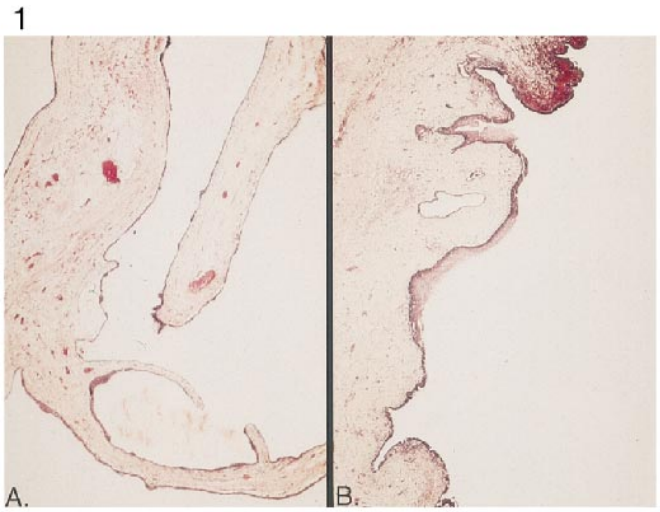
herent fibroadipose tissue. The cysts varied greatly in size and had intervening dense fibrous tissue stroma. The intact cysts were filled with clear to straw-colored fluid or thick mucoid to opaque greenish yellow fluid. One of the cysts in case 2 contained a 1.5-cm solid nodule corresponding to the adenocarcinoma. The histologic features in all cases were quite similar (Figures 1 and 2). A wide variety of lining epithelia were identified in the cysts in each case; stratified squamous epithelium was the most common, seen in all cases, along with cuboidal, transitional, stratified columnar, mucinous or ciliated columnar (3 cases), ciliated pseudostratified columnar (2 cases), and gastric (2 cases) types. In addition, varying numbers of goblet cells were encountered within the columnar lining in 1 case. Two cases had occasional islands of pancreatic acini and islets of Langerhans. These same 2 cases also had occasional foci of mucous and serous glands. Interestingly, all 5 cases had occasional bundles of well-formed smooth muscle fibers separated from the lining epithelium by a thin layer of fibrous tissue; these foci simulated the normal gut wall. The lining epithelium was often eroded, and the underlying stroma showed dense infiltration by inflammatory cells. Skin adnexa, neural elements, and heterologous mesenchymal tissue, such as cartilage and bone, were not identified. The adenocarcinoma in case 2, estimated to be 1.5 cm, was located within a cyst and infiltrated the surrounding stroma. The morphology of this carcinoma was similar to the usual colonic adenocarcinomas with well-formed glands (Figure 3). The carcinoma in case 5, also estimated to be 1.5 cm, was found within the fibrous tissue adjacent to the lining epithelium of a cyst (Figure 4). This carcinoma was composed of multiple solid nests of uniform epithelial cells with a rich capillary network. The tumor cells were occasionally arranged in trabeculae or acini or in single cellular files. Immunostaining for cytokeratin and chromogranin was positive, and electron microscopy revealed many well-formed neurosecretory granules. These findings supported a diagnosis of neuroendocrine carcinoma.

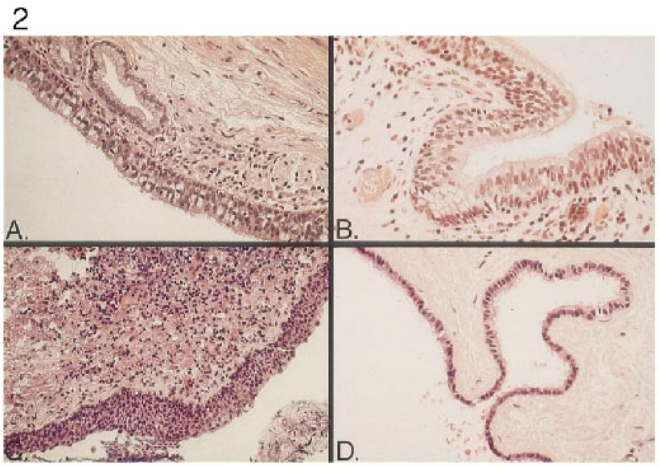
COMMENT

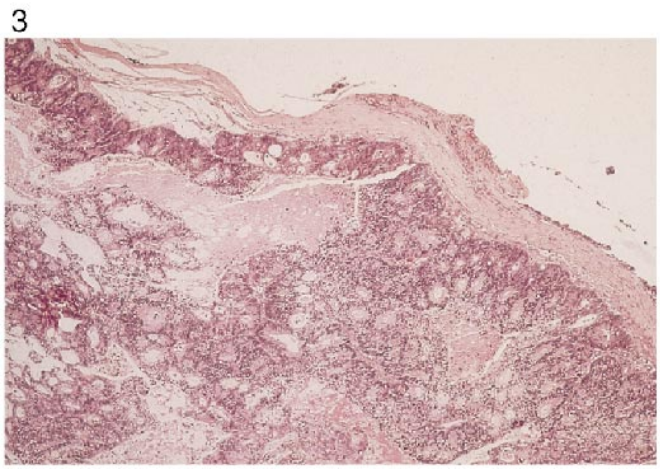
The retrorectal (presacral) space is a potential space bounded anteriorly by the rectum and posteriorly by the sacrum. The peritoneal reflection forms the superior border, and the levator ani and coccygeus muscles form the inferior border. A variety of neoplastic and nonneoplastic conditions occur in this region.[2–6] Teratomas are the more common lesions in children, whereas in adults, chordoma and developmental cysts are more frequent.[1,2,6] Based on morphology, developmental cysts are classified into epidermoid cysts, dermoid cysts, enteric or rectal duplication cysts, retrorectal cystic hamartomas, and cystic teratomas.[7] Epidermoid and dermoid cysts are usually unilocular and are lined by stratified squamous epithelium. Dermal appendages are present in dermoid cysts, but not in epidermoid cysts. Duplication cysts are also unilocular and are lined by epithelium similar to epithelium of the gastrointestinal and respiratory tracts. The epithelium, often with villi, crypts, and glands, simulates the normal mucosa of the gut. The main distinctive feature is a well-formed muscular wall with 2 layers of muscle bundles containing nerve plexuses in between.[8] Retrorectal cystic hamartomas are usually multicystic or multiloculated. The cysts are lined by a wide variety of epithelia, which varies from cyst to cyst or even within the same cyst, including stratified squamous, transitional, stratified columnar, mucinous or ciliated columnar, ciliated pseudostratified columnar, and gastric types. The cyst wall in most cases contains focal, well-formed smooth muscle fibers. However, the muscle bundles are often disorganized and are present focally, unlike the well-formed continuous 2-layer muscle coat seen in duplication cysts.

How these cysts develop is still poorly understood. Are they distinctive or interrelated entities, or do they all represent some form of teratomas? The distinction of retrorectal cystic hamartomas from cystic teratomas is even more difficult, particularly when there is only limited tissue for pathologic examination. Theoretically, retrorectal cystic hamartomas can be classified as teratomas. They possess all 3 germ layers, that is, the ectoderm (squamous), endoderm (intestinal-type epithelium), and mesoderm (smooth muscle & fibrous tissue). However, the term *teratomas* should be reserved for cases with dermal appendages, neural elements, or other heterologous mesenchymal derivatives, such as cartilage and bone. The proper classification of developmental cysts requires thorough sampling of the resected specimens. Many cases could have been misclassified; consequently, it is difficult to study their true incidence.

Retrorectal cystic hamartomas have been described in the literature by various terms, including cyst of postanal intestine, tailgut cyst, mucus-secreting cyst, enterogenous cyst, simple cyst, myoepithelial hamartoma of the rectum, and retrorectal cyst.[1,2,6,9–11] They are presumed to be of developmental origin. Although their precise embryologic basis is unknown, these cysts are believed to arise from







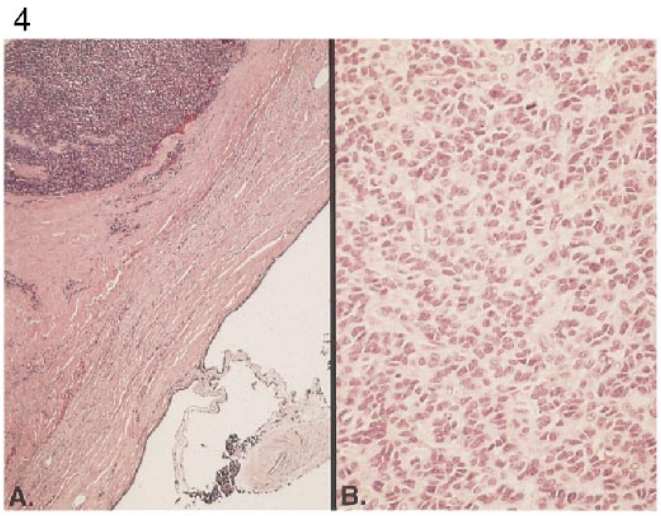


Figure 1. *A, Retrorectal cystic hamartoma: multiloculated cyst (hematoxylin-eosin, original magnification ×10). B, Cyst lining showing transition into different epithelia (hematoxylin-eosin, original magnification ×20).*

Figure 2. *Variety of lining epithelia seen within the cysts (hematoxylin-eosin, original magnification ×100). A, Ciliated pseudostratified columnar epithelium. B, Ciliated stratified columnar epithelium. C, Transitional epithelium. D, Cuboidal epithelium.*

Figure 3. *Adenocarcinoma in a retrorectal cystic hamartoma (hematoxylin-eosin, original magnification ×80).*

Figure 4. *Neuroendocrine carcinoma within the cyst wall. A, Low-power view (hematoxylin-eosin, original magnification ×20). B, High-power view (hematoxylin-eosin, original magnification ×100).*

vestiges of the embryonic hindgut.[1,12] During the 3.5- to 8.0-mm stage of development (approximately 28–35 days' gestational age), the embryo possesses a true tail, and the primitive hindgut extends into this tail, caudal to the site of subsequent formation of the anus.[1,9] This caudal extension is called tailgut or postanal gut. Normally by the eighth week of embryonic development, the tailgut atrophies and the tail involutes. However, sometimes the tailgut fails to regress completely. Some investigators believe that the persistence of such tailgut remnants gives rise to the tailgut cyst, or retrorectal cystic hamartoma.[1,12,13] In 1928, Peyron,[14] in his study of a series of tailgut specimens from mammalian embryos, noted various types of lining epithelium with a predominance of intestinal-type epithelium. A definite muscular or serous coat was lacking in all the specimens.

Retrorectal cystic hamartoma is more commonly reported in middle-aged women,[1,10,15] but it can be detected at any age, including infancy.[1,10,13] It may present as an asymptomatic mass during physical examination or at childbirth.[7,16] If infected, it is often misdiagnosed as a pilonidal cyst, anorectal fistula, or a recurrent retrorectal abscess.[7,17] Discomfort while sitting and rectal bleeding are common symptoms.[6,15] Some of the aforementioned symptoms were also noted in our group of patients, although the diagnosis of a mass lesion was delayed by at least a year in all our cases, suggesting that retrorectal cystic hamartoma is masked by symptoms and signs of other diseases afflicting the anus and anal canal.

Malignant change as a rare complication has been documented occasionally. Ten histologically documented cases have been reported in the literature, including 6 adenocarcinomas and 4 carcinoids (Table 2).[17–25] Two additional reported cases were instances of adenocarcinomas arising in duplication cysts. One case was documented with double layers of smooth muscle in the wall[26]; however, in their review of tailgut cysts, Hjermstad and Helwig regarded this case as a retrorectal cystic hamartoma.[1]

Table 2. Cases of Retrorectal Cystic Hamartomas With Malignancy Reported in the Literature

Case No.	Author, y	Age, y/ Sex	Clinical Features	Gross Features	Malignancy	Treatment and Follow-up
1	Ballantyne,[18] 1932	38/F	Discomfort while sitting; deficiency of lower end of sacrum on radiograph	Unilocular cyst	Adenocarcinoma	DOD 7 mo postoperatively with recurrence and metastasis to groin lymph nodes and lung
2	Marco et al,[19] 1982	62/F	Discomfort on sitting; mass since childhood; barium enema normal	Multicystic mass; 15 × 8.5 × 6 cm with 3–4-mm granular area	Adenocarcinoma	ANED 20 mo postoperatively
3	Hjermstad,[20] 1985	31/F	Presacral swelling and tenderness; continuous pain left buttock; calcifications on radiograph	Multilocular cyst, 10 cm	Adenocarcinoma	DOD 8 mo postoperatively
4	Hood et al,[21] 1988	N/A	Constipation; prerectal mass on digital examination	Multilocular cyst with solid areas and thick green fluid contents	Carcinoid	N/A
5	Hood et al,[21] 1988	50/F	Constipation; rectrorectal mass on digital examination; complex solid and cystic mass on CT scan	Multilocular cysts with solid areas and thick green fluid contents	Carcinoid	N/A
6	Lin et al,[22] 1992	18/F	Perianal pain and difficulty in urination; cyst with 1 solid area on CT scan	Multilocular cyst, 10 × 6 × 5 cm, with solid protruding mass in 1 locule	Carcinoid	N/A
7	Liessi et al,[23] 1995	50/M	Presacral mass discovered on digital examination and CT scan	N/A	Adenocarcinoma	Local recurrence 6 mo postoperatively; final outcome N/A
8	Levert et al,[17] 1996	63/F	Discomfort on sitting; 9 cm mass on CT scan	Multilocular cyst, 6 × 5 cm	Adenocarcinoma	ANED 5 y postoperatively
9	Horenstein et al,[24] 1998	19/F	Pelvic pain; irregular menstrual cycle; cystic mass on ultrasound	Multicystic mass, 8 cm	Carcinoid	ANED 4 y
10	Lim et al,[25] 1998	40/F	Urinary frequency; constipation; cyst with fluid and localized thickening of wall on MRI	25 × 10 × 10 cm with dense mucoid contents	Adenocarcinoma	ANED
11	Present report	36/F	Asymptomatic; retrorectal mass on digital examination during routine physical	9.5 × 9.2 × 8.8-cm multilocular cystic mass with 3 × 2 × 2.8-cm solid area	Adenocarcinoma	ANED 2 y postoperatively
12	Present report	69/F	Mild rectal bleeding and pain with bowel movements	4-cm presacral cystic mass on CT scan	Neuroendocrine carcinoma	ANED 2 y postoperatively

* N/A indicates not available; CT scan, computed tomographic scan; MRI, magnetic resonance imaging; DOD, died of disease; and ANED, alive with no evidence of disease.

The other case was not well documented histologically as a duplication cyst.[27] The strong association of carcinoid tumors and retrorectal cystic hamartomas (4 out of 10 cases in the literature and 1 of our cases with a neuroendocrine carcinoma) suggest the possibility that some of the presacral carcinoid tumors reported in the literature may arise from retrorectal cystic hamartomas. The possible relationship of presacral carcinoid tumors with tailgut cysts has also been alluded to by Horenstein et al[24] and others.[28–30]

The clinical significance of retrorectal cystic hamartomas mainly concerns the morbidity that can result if the lesion is not suspected and definitive surgery is not undertaken. The potential for infection, occurrence of recurrent perianal fistulas, and the possibility of malignant transformation emphasize the importance of early complete surgical excision of these lesions.

In conclusion, retrorectal cystic hamartoma appears to be a distinct clinicopathologic entity occurring most commonly in young adult women. The anatomic location and the variety of epithelia seen in retrorectal cystic hamartoma support its origin from the tailgut vestiges. The clinical diagnosis of retrorectal cystic hamartomas is often delayed, partly due to unfamiliarity with this entity and also due to its symptomatological mimicry of other more commonly occurring lesions at this site, including perianal fistulas and abscesses. The pathologic diagnosis based on biopsy is also difficult. The biopsy specimens often contain only inflamed fibrous tissue without epithelia or only 1 type of epithelium, usually squamous. In the latter situation, it may be difficult to distinguish retrorectal cystic hamartoma from other types of developmental cysts. Retrorectal cystic hamartomas should be suspected in women with multiple recurrences of anal fistula. Malignancy, although unusual, does occur in retrorectal cystic hamartomas and may be focal; meticulous gross examination and thorough sampling are therefore important.

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