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 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 excision 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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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-
inferior border. A variety of neoplastic and nonneoplastic der, and the levator ani and coccygeus muscles form the sacrum. The peritoneal re¯ection forms the superior bor-

ed a diagnosis of neuroendocrine carcinoma. Immunostaining for cytokeratin and chromogranin was formed neurosecretory granules. These ®ndings support-

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of a cyst (Figure 4). This carcinoma was composed of mul-

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moma in case 5, also estimated to be 1.5 cm, was found

heterologous mesenchymal tissue, such as cartilage and

broadipose tissue. The cysts varied greatly in size

and had intervening dense fibrous tissue stroma. The in-
tact cysts were ®lled with clear to straw-colored ¯uid or

thick mucoid to opaque greenish yellow ¯uid. One of the

cysts in case 2 contained a 1.5 cm solid nodule corre-

sponding to the adenocarcinoma. The histologic features in

all cases were quite similar (Figures 1 and 2). A wide

variety of lining epithelia were identi®ed in the cysts in

each case; stratified squamous epithelium was the most

common, seen in all cases, along with cuboidal, transi-
tional, stratified columnar, mucinous or ciliated columnar

(3 cases), ciliated pseudostrati®ed 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. Inter-
estingly, all 5 cases had occasional bundles of well-formed

smooth muscle ®bers separated from the lining epitheli-

um by a thin layer of ®brous tissue; these foci simulated

the normal gut wall. The lining epithelium was often eroded,

and the underlying stroma showed dense ®ltration

by in®ammatory cells. Skin adnexa, neural elements, and

heterologous mesenchymal tissue, such as cartilage and

bone, were not identi®ed. 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 adenocar-
cinomas with well-formed glands (Figure 3). The carci-

noma in case 5, also estimated to be 1.5 cm, was found

within the ®brous tissue adjacent to the lining epithelium

of a cyst (Figure 4). This carcinoma was composed of mul-
tiple solid nests of uniform epithelial cells with a rich cap-

illary network. The tumor cells were occasionally ar-
nanged in trabeculae or acini or in single cellular ®les.

Immunostaining for cytokeratin and chromogranin was

positive, and electron microscopy revealed many well-

formed neurosecretory granules. These ®ndings support-
ed 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 re®ection forms the superior bor-
der, 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.3,26 Based on
morphology, developmental cysts are classi®ed into epi-
dermoid 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 ap-
pendages are present in dermoid cysts, but not in epider-
moid cysts. Duplication cysts are also unilocular and are
lined by epithelium similar to epithelium of the gastro-
intestinal and respiratory tract. The epithelium, often
with villi, crypts, and glands, simulates the normal mu-
cosa of the gut. The main distinctive feature is a well-
formed muscular wall with 2 layers of muscle bundles
containing nerve plexuses in between.5 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 pseudostrati®ed

columnar, and gastric types. The cyst wall in most cases
contains focal, well-formed smooth muscle ®bers. How-
ever, 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 rep-
resent some form of teratomas? The distinction of retro-
rectal cystic hamartomas from cystic teratomas is even
more dif®cult, particularly when there is only limited tis-
sue for pathologic examination. Theoretically, retrorectal
cystic hamartomas can be classi®ed as teratomas. They
possess all 3 germ layers, that is, the ectoderm (squau-
mous), endoderm (intestinal-type epithelium), and meso-
derm (smooth muscle & ®brous tissue). However, the term
teratomas should be reserved for cases with dermal ap-
pendages, neural elements, or other heterologous mesen-
chymal derivatives, such as cartilage and bone. The proper
classi®cation of developmental cysts requires thorough
sampling of the resected specimens. Many cases could
have been misclassi®ed; consequently, it is dif®cult 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, entrerogenous
cyst, simple cyst, myoepithelial hamartoma of the rectum,
and retrorectal cyst.1,2,6–11 They are presumed to be of de-
velopmental origin. Although their precise embryologic
basis is unknown, these cysts are believed to arise from

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

* CT indicates computed tomographic scan; MRI, magnetic resonance imaging; and ANED, alive with no evidence of disease.
vestiges of the embryonic hindgut. 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. 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. In 1928, Peyron, 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, but it can be detected at any age, including infancy. It may present as an asymptomatic mass during physical examination or at childbirth. If infected, it is often misdiagnosed as a pilonidal cyst, anorectal fistula, or a recurrent retrorectal abscess. Discomfort while sitting and rectal bleeding are common symptoms. 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). 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; however, in their review of tailgut cysts, Hjermstad and Hélwig regarded this case as a retrorectal cystic hamartoma.
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 al24 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.

References

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

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

* 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.