Retrorectal epidermoid cyst with unusually elevated serum SCC level, initially diagnosed as an ovarian tumor

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Abstract

Retrorectal epidermoid cyst is one of the developmental cysts which arise from remnants of embryonic tissues. We report a rare case of retrorectal epidermoid cyst, initially diagnosed as an ovarian tumor. Serum SCC value as tumor marker was elevated to the high level. Laparoscopy revealed ovaries, uterus and other pelvic organs were all normal. This tumor existed in the retroperitoneal cavity and compressed the rectum. Later, complete tumor resection was performed by laparotomy. Histological study revealed the epithelium of this tumor consisted of only squamous cells without atypia, and the diagnosis of this tumor was retrorectal epidermoid cyst. Retrorectal epidermoid cyst is very rare, and difficult to diagnose before surgery. However, if we have knowledge of developmental cysts, and by careful digital examination and image diagnosis, a differential diagnosis can be made.

Introduction

Retrorectal epidermoid cyst is one of the developmental cysts which arise from remnants of embryonic tissues. Because of the rarity of these tumors in adults, the differential diagnosis is very difficult before surgery. We report a rare case of retrorectal epidermoid cyst which showed the elevated serum SCC level, and was initially diagnosed as an ovarian tumor.

Case Report

A 22-year old woman, gravida 1, para 0, presented with urinary discomfort and constipation from one month. She had not been able to urinate for a few days previously without pressing herself. She consulted a previous hospital's department of obstetrics and gynecology. In that hospital, the diagnosis was an ovarian tumor, and she was introduced to our department for curative surgery. In the internal and rectal examination, a hard, cystic, non-removable, mass larger than a fist was palpated in the left side of rectum and the posterior of the uterus. We could not insert a speculum and a probe for transvaginal sonography into her vagina because of the tumor's strong compression. MRI revealed a unilocular cystic tumor measuring 11x11x12 cm in the bottom of the pelvic cavity, compressed rectum, uterine cervix, vagina and urinary bladder (Figure 1). The contents of this cyst consisted of two different kinds of fluid, and there was a borderline between them. The content in the back side had T1 weighted-low intensity, T2 weighted-high intensity signal. The abdominal side showed T1 weighted-iso intensity, T2 weighted-moderately high intensity signal. There were some heterogeneous contents in both fluids (Figure 1). Given the fat suppressive condition, this T1 weighted-iso intensity area slightly reduced its signal. The content in the center of the abdominal fluid had much reduced its signal by this condition. This means the abdominal side of this cyst contains a fat element. The wall in this cyst was thin and smooth. There was neither solid part in the tumor nor signs of invasion into surrounding tissues. White blood cell (10.5×10^9/L) and C-reactive protein (2.05 mg/dL) were slightly elevated. Other laboratory data were within normal limits. In these clinical findings, the diagnosis before the operation was an ovarian tumor, and in particular, a mature cystic teratoma of the ovary was suspected from image diagnosis. Laparoscopy was performed for diagnosis and treatment in the same day. However, it revealed ovaries, uterus and other intra-peritoneal pelvic organs were all normal. This tumor was in the retroperitoneal cavity, and compressed the rectum to the right and anterior side (Figure 2). Since we could not make a diagnosis of this retroperitoneal tumor, we consulted the department of surgical gastroenterology, and we abandoned the sequential curative operation by laparoscopy. More clinical examinations were performed after laparoscopy including colonoscope, 3D CT. Serum tumor markers were also proven out after laparoscopy. SCC value was elevated to 30.4 ng/mL (cut-off value is 1.5 ng/mL). Other tumor markers such as CA125, CA19-9, CEA, AFP, CA72-4, CA15-3 and TPA were all within normal limits. On the following days, laparotomy was performed for curative excision of this tumor with some malignant possibilities because of the elevated serum SCC level. This tumor adhered to the posterior wall of the rectum, but complete resection was possible and was performed. Both intra-operative frozen section analysis and the cytology of the tumor's liquid contents did not show any malignant finding. The resected-tumor measured 12×10 cm, and it was light gray-colored, elastic, hard and unicystic. The cyst wall was thin and degenerated. It weighed approximately 500 grams as a whole tumor. The content of this cyst was filled with 300 mL of dense muddy and fatty fluid, which partially curdled.

The cytology of this content showed abundant white blood cells and squamous cells without any atypia (Figure 3). Histological study revealed that this tumor had a unicystic structure, and the epithelium of this cyst wall consisted of only keratinized, stratified squamous cells (Figure 4A and B). Some portion of squamous epithelium almost maintained a stratified layer structure (Figure 4A), another portion of these peeled off from their outer surface (Figure 4B). There was no atypia of squamous epithelium. According to these pathological findings, this tumor was diagnosed as an epidermoid cyst, which was one of

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Contributions: MH, mainly took charge of this patient as a gynecologist, and conceived, designed and supervised this manuscript; AS, KT and NK, also took charge of this patient; IF and NI, reviewed this manuscript; ST and TF, diagnosed this tumor pathologically,and performed immunohistochemical study; HN and KK, took charge of this patient as a surgeon.

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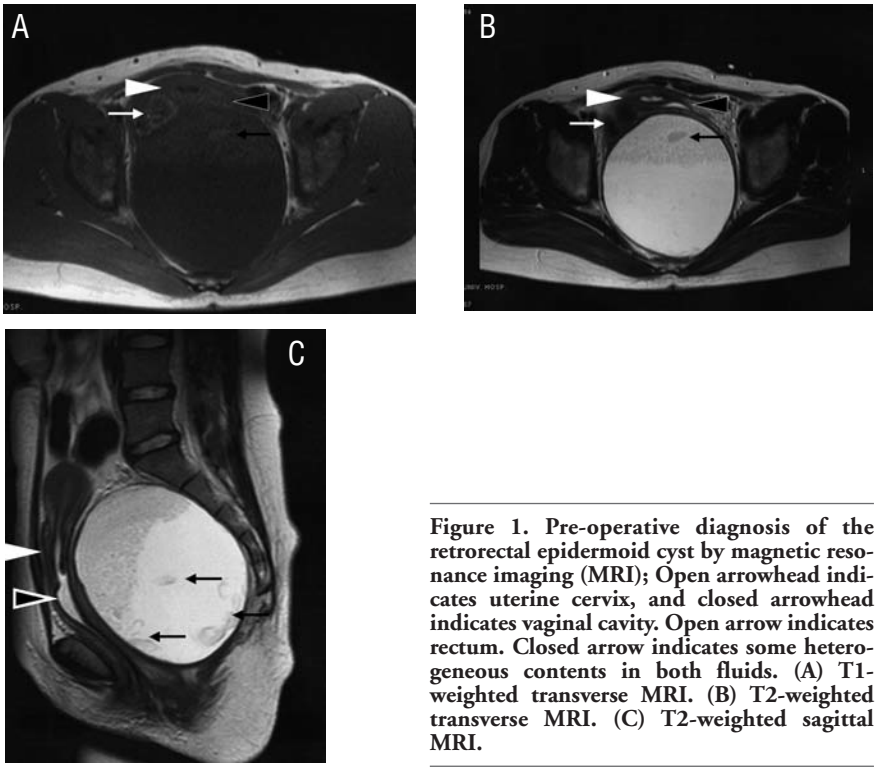


Figure 1. Pre-operative diagnosis of the retrorectal epidermoid cyst by magnetic resonance imaging (MRI); Open arrowhead indicates uterine cervix, and closed arrowhead indicates vaginal cavity. Open arrow indicates rectum. Closed arrow indicates some heterogeneous contents in both fluids. (A) T1-weighted transverse MRI. (B) T2-weighted transverse MRI. (C) T2-weighted sagittal MRI.

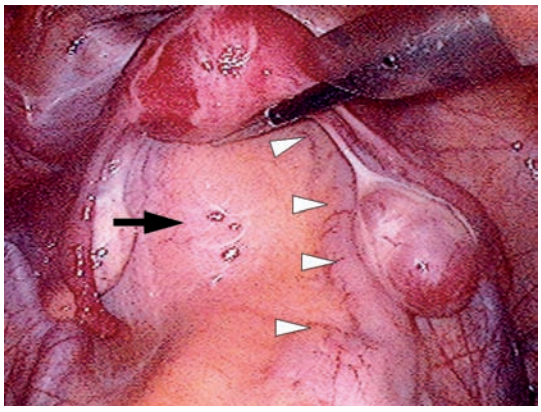


Figure 2. Laparoscopic finding in the pelvic cavity; open arrowhead indicates rectum, which was compressed and shifted to the right and anterior side by the retrorectal epidermoid cyst. Arrow indicates the retrorectal epidermoid cyst as a retroperitoneal tumor.

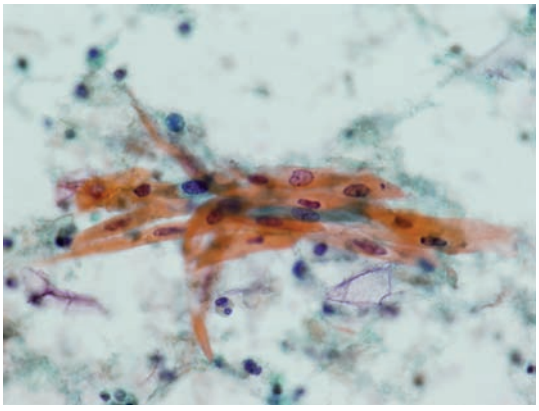


Figure 3. Cytology of the contents in the presacral epidermoid cyst; Papanicolaou stain (x800).

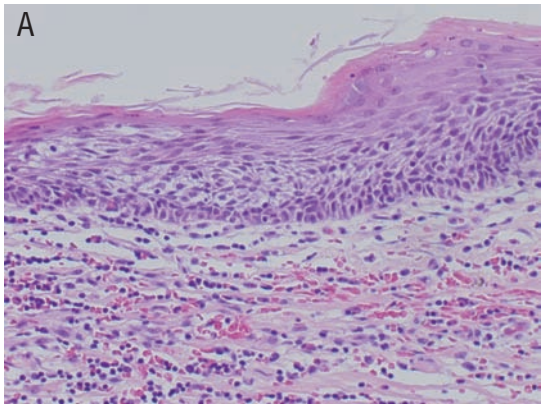
the developmental cysts. Immunohistochemical studies using anti-CEA, CA19-9, and SCC-antigen antibodies were performed. CA19-9 was negative. On the other hand, CEA was weekly positive in the stratified squamous epithelium (Figure 5A1) and moderately positive in the peeled and peeling keratinized epithelium (Figure 5A2). SCC-antigen was strongly positive in the stratified squamous epithelium (Figure 5B1). Especially in the peeled and peeling keratinized epithelium, it was intensively positive (Figure 5B2). After resection of this tumor, serum SCC level was normalized. This case has been free of recurrence and her serum SCC level also has remained within normal range for two years after surgery.

Materials and Methods

Immunohistochemical study was performed by the common avidin-biotin complex method using anti-CEA antibody (Histofine SAB-PO(M) Kit®; Nichirei Bio Science, Japan), anti-CA19-9 antibody (DAKO, USA) and anti-SCC-antigen antibody (Hepa-Ab®; XEPTAGEN, Italy). Hepa-Ab® recently became available for the detection of SCC-antigen in the immunohistochemical study.

Discussion

Retrorectal cysts are rare in adults. The incidence of these tumors is estimated at 1/40,000-63,000 admissions to large referral centers.[1] The etiology of these cysts is classified into five categories,[2] which are congenital, neurogenic, inflammatory, osseous and miscellaneous. Most cases of 16 retrorectal cysts are congenital for 55-70%,[1-3] and most of congenital retrorectal cysts are developmental cysts, which are 60% of them.[1,2] The term "developmental cysts" was initially proposed and defined by Hawkins in 1953.[4] These are thought to arise from caudal embryonic vestiges,[4] and occur mostly in middle-aged women and in 3:1 female:male ratio.[5] Originally, these are classified into the three subtypes by histological findings, which are epidermoid cyst, dermoid cyst, and tailgut cyst.[4] Both epidermoid cyst and dermoid cyst are lined with stratified squamous epithelium, but dermoid cyst also contains skin appendages, such as hair follicle, sweat glands and tooth buds. Both cysts result from closure defects of the ectodermal tube with skin inclusion. Tailgut cyst, also known as retrorectal cystic hamartoma or mucus secreting cyst, is lined with various epitheliums such as columnar cells, squamous cells, and transitional cells, often in combinations of these. This cyst arises from the remnants of the embryonic postnatal gut.[4-7] The clinical presentation of retrorectal development cysts is often non-specific, and half of them are asymptomatic, which are incidentally discovered during routine physical examination. The most common symptom is related to tumor effect arisen by compression to the rectum and lower urinary tract, such as constipation and urinary frequency.[5-9] The other common symptom is pain when these cysts are infected or generate malignant transforma-



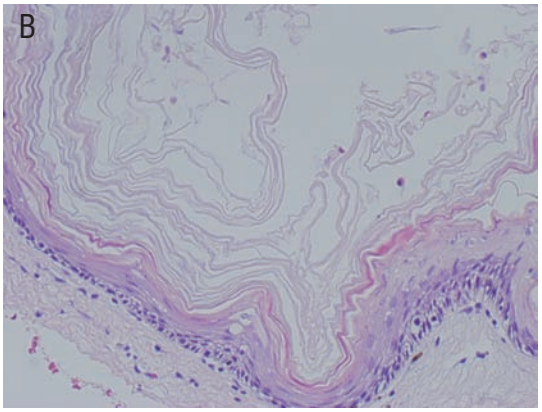
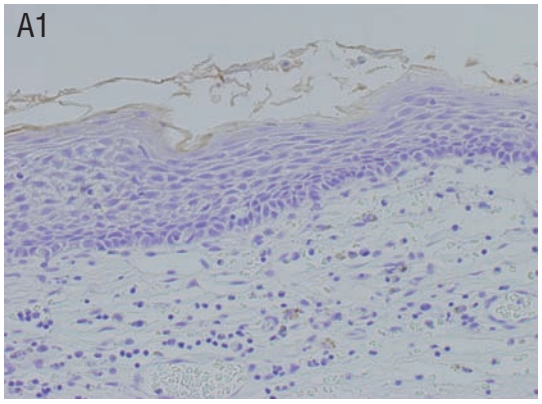
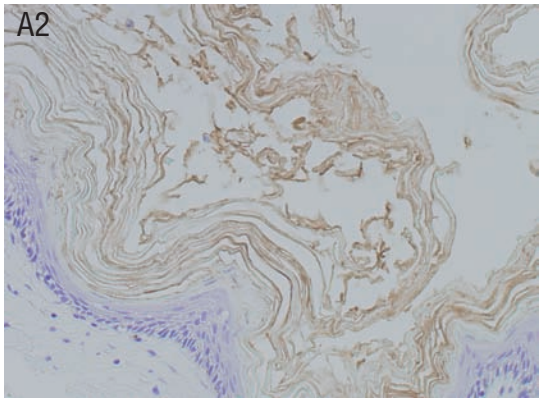
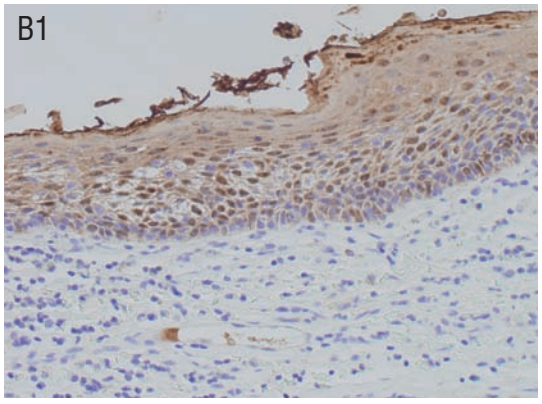


Figure 4. Pathological features of the retrorectal epidermoid cyst; H.E. stain (x200). (A) Stratified squamous epithelium. (B) Peeling and peeled squamous epithelium.







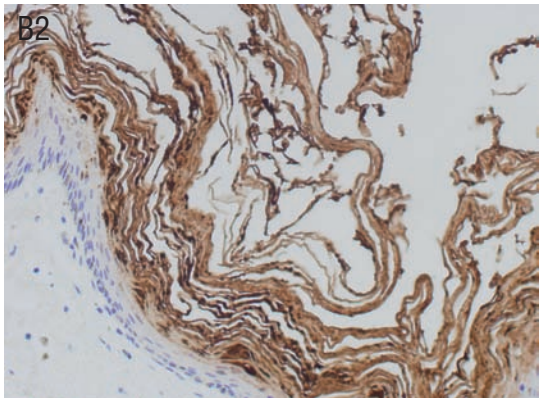


Figure 5. Immunohistochemical studies of the retrorectal epidermoid cyst. (A) Immunostaining for CEA (x200). (A1) Stratified squamous epithelium. (A2) Peeling and peeled squamous epithelium. (B) Immuno-staining for SCC antigen (x200). (B1) Stratified squamous epithelium. (B2). Peeling and peeled squamous epithelium.

tion.[6,9] Chronic infection occurs in 30-50% of developmental cysts.[10] Infected cysts are source of a perianal abscess or a draining sinus. How infection originates is unknown, but trauma and infection by the lymphatics or a congenital sinus have been suggested.[7] Malignant transformation of developmental cysts is rare. Abel reported an incidence of 8% in developmental cysts.[10] Hjermstad reported one case of malignant transformation, which histology confirmed was poorly differentiated mucinous adenocarcinoma, in 53 cases of tailgut cysts.[11] Killingstone reviewed 43 cases of tailgut cyst. The malignancy arose in 17 cases of these; adenocarcinoma in 11 cases, carcinoid in 5 cases and neuroendocrine in one case.[12] In Japan, 58 cases of epidermoid cysts have been reported including our case. Malignant transformation occurred in 4 cases (6.9%) of these; all were squamous cell carcinoma.[13] For the treatment of developmental cysts, complete surgical resection is recommended so as not to cause infection, recurrence or generate malignant transformation.[5,7,9] If the patients do not have any clinical[17] symptom or sufficient clinical findings present any possibilities of malignancy, surgical resection should be performed because of the difficulties of operating once infection has occurred in the developmental cysts.[7] Regarding serum tumor markers of the developmental cysts, there were some reports that serum CEA and/or CA19-9 level were elevated in the malignant tailgut cases.[14-16] However, in the retrorectal epidermoid cyst, there is no report that the serum tumor marker is positive as far as we know in the English language literature. Nagano reported SCC, CA19-9, CA125 and CEA levels in the aspirated fluid of one malignant retrorectal epidermoid cyst case (squamous cell carcinoma) were all extensively elevated; however, all serum levels were within the normal limit. We did not measure the tumor marker level of its contents. From the result of immunohistochemical study and this case's clinical course, we supposed elevated serum SCC level was the cause in this case. Subacute inflammation on her epidermoid cyst occurred due to physical stimuli or unknown cause, and then squamous cell of epithelium in the epidermoid cyst degenerated and peeled off. Consequently, the SCC-antigen

was released into the blood circulation. Because of the rarity of developmental cysts, they are very frequently misdiagnosed and inappropriate surgery is performed.[6] If a gynecologist initially finds a retrorectal cyst, most cases will be misdiagnosed as an ovarian tumor. Retrorectal epidermoid cyst contains fatty elements such as desquamated debris, cholesterol, keratin, and water.[18] A gynecologist confuses these elements in the epidermoid cyst with that of mature cystic teratoma, which is a common ovarian tumor. However, if we have knowledge of the developmental cysts, by careful digital examination and image diagnosis, it is possible to make a differential diagnosis, since developmental cysts exist between the presacral and retrorectal space, not in the Douglas pouch like an ovarian tumor.[18]

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