Case History:

A 56 year-old woman with a history of irritable bowel syndrome presented to a walk-in care center with acute pelvic pain and pressure. Computed tomography (CT scan) then demonstrated a 3-cm-ovoid mass at the right retrorectal space. CT-guided core biopsy was performed, but the tissue obtained was non-diagnostic.

On exam, digital rectal exam revealed a small rectocele and a soft, mobile mass posterior and to the right of midline.

A subsequent MRI was performed, which showed a multilocular cystic mass adjacent to and displacing an otherwise normal rectum (Figure A).

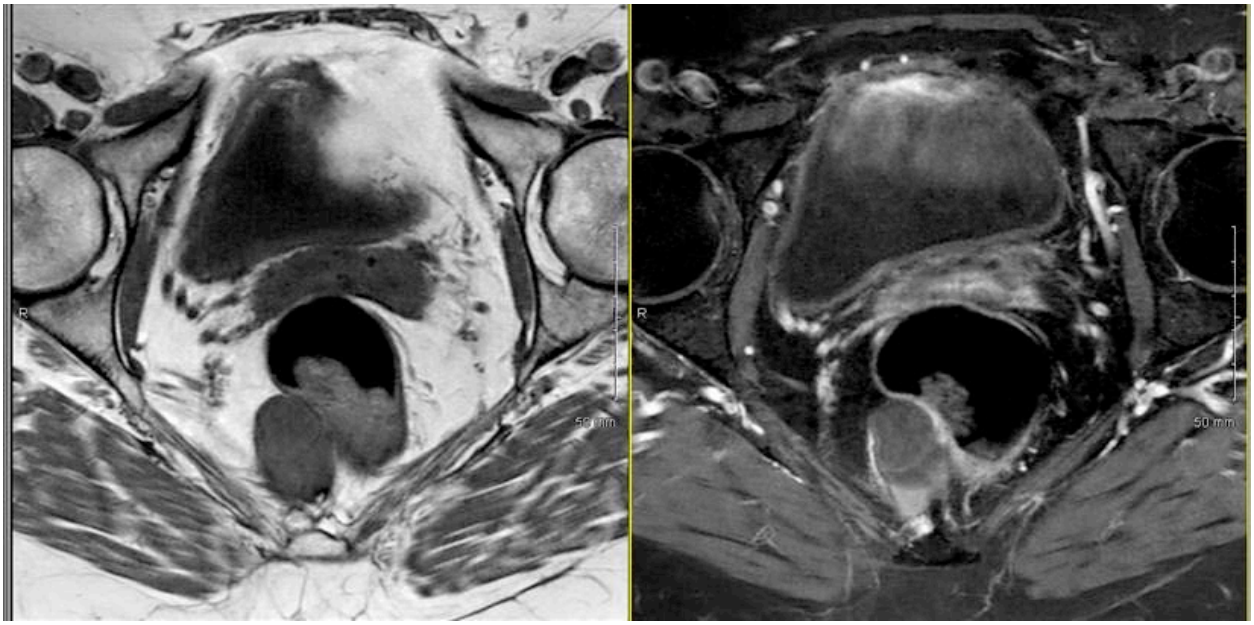

Figure A: T1–weighted MR images with (right) and without (left) fat saturation demonstrate a multilocular, cystic mass adjacent to and displacing the otherwise normal rectum. No solid enhancing components were identified.

A surgical excision was performed; pathology received a multilocular, smooth-lined cyst weighing 5.2 grams and measuring 3.7 x 2.2 x 1.5 cm. It contained clear fluid as well as soft yellow material. On microscopic examination of the entire submitted specimen, the cyst was lined by bland columnar epithelium (H&E figures B-C) intermixed with squamoid and transitional foci (H&E figures C- D). Margins of resection were negative for lesional tissue.

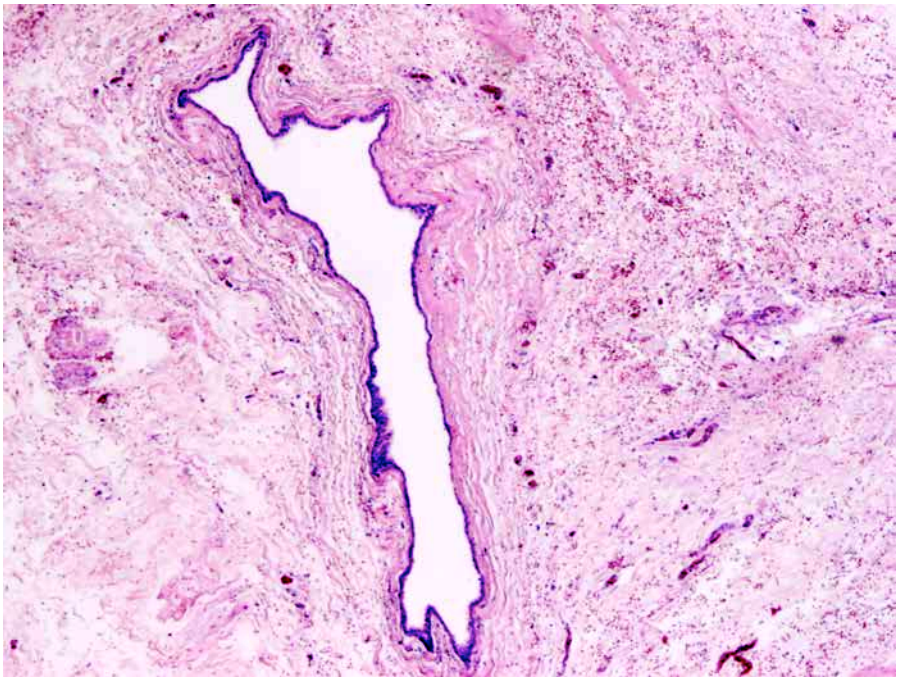

Figure B: Cyst with hypocellular fibro-connective wall and mild chronic inflammation (H&E -40x magnification)

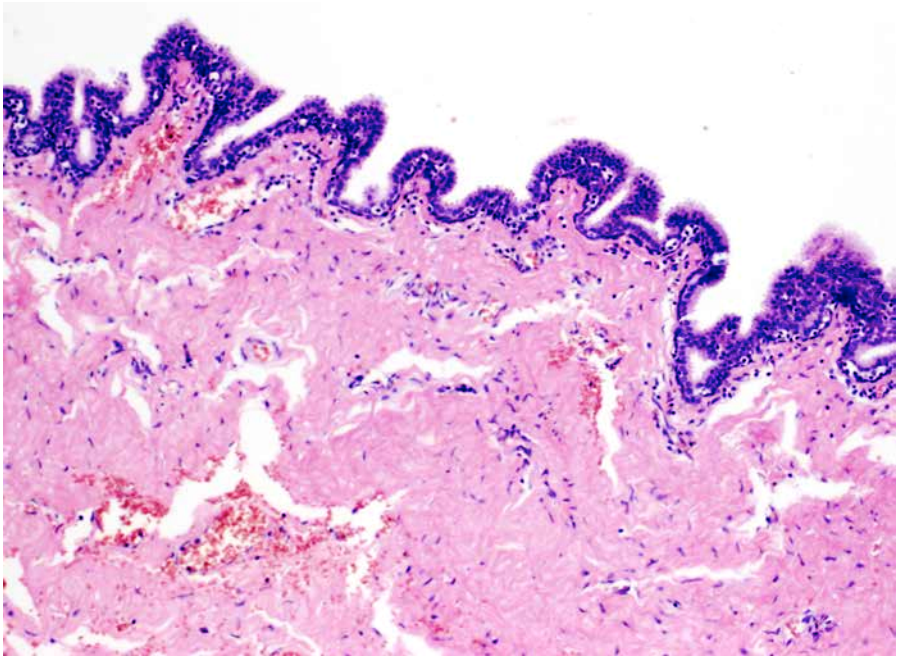

Figure C: Benign simple and stratified columnar epithelium (H&E- 100x magnification)

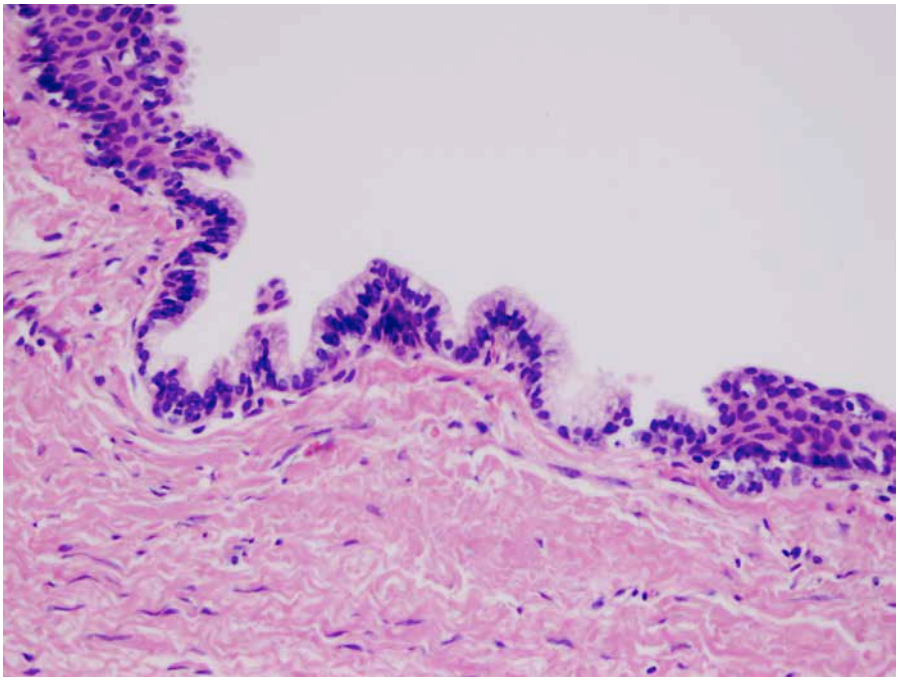

Figure D: Variation in lining with intermix of squamoid and columnar epithelium, negative for dysplasia (H&E- 200x magnification)

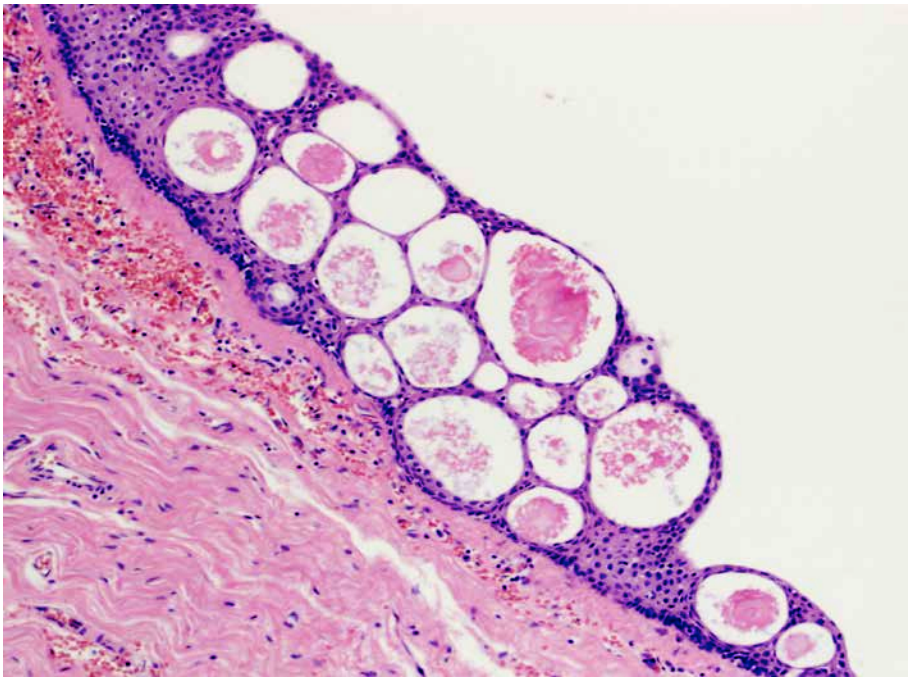

Figure E: Transitional-type epithelium with cystic change, resembling changes of cystitis cystica (H&E- 100x magnification)

Based on descriptions and histological imaging provided above what is your diagnosis?

A. Enteric/rectal duplication cyst

B. Tailgut cyst

C. Epidermoid/Dermoid cyst

D. Müllerian Cyst

E. Teratoma

Answer: (B) Tailgut cyst

Discussion

The retrorectal space is the potential space bounded by the rectum anteriorly, the sacrum posteriorly, the peritoneal reflection superiorly, and the levators ani and coccygeus muscles inferiorly. The ureters and iliac vessels are the lateral margins. Although many tailgut cysts are asymptomatic, classic presenting symptoms include those related to compressive/ pressure effect such as chronic perirectal pain, rectal fullness and urinary frequency or may present with change in stool pattern and painless rectal bleeding.[1, 2]

Tailgut cysts are rare congenital lesions thought to arise from the remnants of the embryonic postanal gut. During embryogenesis, humans develop a true tail at 28-35 days of gestation, and the embryonic hindgut extends into this tail, forming the tailgut (or postanal gut). The tail, along with the tailgut, undergoes regression by 56 days gestational age. It is theorized that tailgut cysts result from failure of complete regression. They most commonly present in middle aged women. [1, 2]

Tailgut cysts are also known as retrorectal cystic hamartomas or tailgut vestige. Grossly, they are unilocular or more commonly multilocular cystic structures filled with fluid which varies from clear, thin and colorless to yellow-green or even opaque brown pasty material. These cysts average 3-4 cm and are circumscribed but not unencapsulated, larger cysts up to 30 cm been reported.[3] (Figure F) Histologically, the cyst lining can be a number of epithelial types including stratified squamous, transitional, mucinous, ciliated columnar or cuboidal mucus secreting cells. Based on AFIP review criteria, cyst must be lined at least in part by columnar or transitional epithelium and lack myenteric plexus or serosa [1, 2].

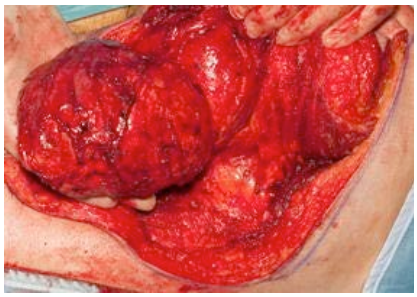

Figure F- In situ image of a large Tailgut cyst (Stallard *et. al*[3])

Malignant transformation is reported in about 13% of tail gut cysts. No specific clinical variable (patient age, mass size, duration of symptoms, or detection on digital rectal examination) can predict malignant transformation. Reported malignancies arising from the tail gut cysts include adenocarcinoma, carcinoid tumor, neuroendocrine carcinoma, squamous, or adenosquamous carcinoma.[4, 5] This association further stresses the need for complete submission of the cyst to exclude teratomatous and/ or malignant components.

Because of the risk of malignant transformation, fistula tract formation, infection, and recurrence, tailgut cysts should be completely excised with regular post-operative follow-up. Although pre-operative presacral biopsy may be indicated when a potential tailgut cyst has a mixed cystic and solid appearance on imaging to evaluate for malignant transformation, biopsy is not indicated in a purely cystic lesion. In fact, biopsy of tailgut cysts, especially transrectal or transvaginal biopsy, evokes concern for infection and seeding.[4, 5]

The differential for a cystic mass in the retrorectal (presacral) space is broad and additionally includes inflammatory and metastatic processes, mullerian cysts, cystic lymphagiomas, chordomas, anterior sacral meningoceles, and other developmental cysts such as teratoma, dermoid, epidermoid, and enteric duplication cysts.

Rectal duplication cysts (Choice A), unlike tailgut cysts, are usually unilocular and composed of gastrointestinal columnar type lining (often with crypts, villi or glands) surrounded by two well-formed layers of smooth muscle with nerve plexuses. In comparison, tailgut cysts have disorganized smooth muscle within the cysts wall and do not contain neural plexuses.[2]

Epidermoid/dermoid cysts (Choice C) classically have a simple cystic structure with keratin debris. Microscopically, the cysts are lined by stratified squamous epithelium. Epidermoid cysts are lined by squamous epithelium only, while dermoid cysts include dermal appendages (pilosebaceous units and/or apocrine sweat glands) in addition to squamous epithelium.

Mature teratomas (Choice E) of the sacrococcygeal region, like tailgut cysts are usually multilocular. Histologically, teratomas contain cells originating from all three germ cell layers: ectoderm, endoderm, and mesoderm, most commonly in the form of dermal appendages, neural tissue, bone or cartilage. Unlike tailgut cysts, mature teratomas frequently demonstrate intralesional calcifications, fat and bony destruction of the coccyx or sacrum. [2, 5]

Müllerian cysts (choice D) or paramesonephric cysts are thought to be Müllerian duct remnants. They normally present paravertebrally, but can be located anywhere, including in mesentery or retroperitoneum. These cysts are lined by simple cuboidal or columnar, non-mucinous and often ciliated epithelium, resembling uterine- tubal epithelium. Peg-cells can be seen, and their lining is generally positive for CK7, estrogen and progesterone receptors by immunoproxidases. Müllerian cysts are often asymptomatic, but rarely associated with pain and expanding size, hypothesized attributable to hormonal influence.[6, 7]

While meningoceles, chordomas and cystic lymphangioma may present similarly by clinical means, they are histologically distinct from tailgut cyst. Menigoceles communicate with the meninges. They contain clear fluid and thick fibrous wall lined by flattened arachnoid cells with variable neural tissue and calcification. Chordomas classically present as a heterogenous sacral mass with evidence of bone destruction and extension into the soft tissue. They form into cord-like arrangement of cells, particularly the large vacuolated physaliferous cells in a background of mucoid tissue and fibrous septa. Cystic lymphangiomas contain large irregular vascular spaces lined by bland endothelial cells and containing proteinaceous material (lymph).

References:

1. Hjermstad BM, Helwig EB. Tailgut cysts. Report of 53 cases. *Am J Clin Pathol*. 1988; 89:139-147.

2. Killingsworth C and Gadacz TR. Tailgut Cyst (Retrorectal Cystic Hamartoma): Report of a Case and Review of the Literature. *Am Surg*. 2005; 71: 666-673.

3. Stallard J, Davies J, Bourke G, Finan PJ. Successful removal of a massive retrorectal cystic hamartoma (tailgut cyst) using a combined paracoccygeal and myocutaneous gluteal flap approach. *Tech Coloproctol*. 2014 Feb 6. [Epub ahead of print]

4. Mathis KL, Dozois EJ, Grewal MS, Metzger P, Larson DW, and Devine RM. Malignant risk and surgical outcomes of presacral tailgut cysts. *British Journal of Surgery*. 2010; 97: 575-579.

5. Chhabra S, Wise S, Maloney-Patel N, Rezac C, Poplin E. Adenocarcinoma associated with tail gut cyst. *J Gastrointest Oncol*. 2013; 4: 97-100.

6. Lee J, Song S, Park CS and Kim B. Mullerian cysts of the mesentery and retroperitoneum: A case report and literature review. *Pathology International*. 1998; 48: 902-906.

7. Boghossian S, Dasgupta K, Haugk B, and White S. Mullerian cyst of the mesentery: A case report of an unusual location. *Clinical Anatomy*. 2010; 23: 866-869.

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