

Atypical Transitional Cell/Squamoid Carcinoma Arising from a Tailgut Cyst

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Abstract

A 59-year-old woman was found to have a 4 cm multi-lobulated mass arising from the posterolateral rectal wall with an adjacent prominent mesorectal lymph node. Correlative CT, MRI and FDG-PET studies were performed, with intense FDG avidity in the rectal lesion and FDG uptake in the lymph node. Core biopsy favoured an atypical transitional cell/squamoid carcinoma, probably arising from a tailgut cyst. The patient was treated with definitive chemoradiotherapy with no evidence of residual/recurrent disease on repeat FDG-PET.

Keywords

Tailgut cyst, Carcinoma, FDG-PET, Transitional cell, Squamoid

Abbreviations

MRI: Magnetic Resonance Imaging; **CT:** Computed Tomography; **PET:** Positron Emission Tomography; **FDG:** 18-Fluorine-Fluorodeoxyglucose; **SUVmax:** Maximum Standardised Uptake Value

Case

A 59-year-old woman was referred for right hip magnetic resonance imaging (MRI) for right hip pain. An incidental 40 mm x 31 mm x 38 mm multi-lobulated mass was found arising from the right posterolateral rectal wall with homogeneous Gadolinium-enhancing internal septations, cystic components and an adjacent enhancing 8 mm mesorectal lymph node (Figure 1). She was referred for surgical review with a firm mass felt 5 cm from the anal verge but no mucosal lesion on colonoscopy. She had no significant medical history and was asymptomatic of the rectal lesion.

Correlative imaging with positron emission tomography (PET) and concurrent diagnostic quality computed tomography (CT) demonstrated intense 18-Fluorine-fluorodeoxyglucose (FDG) uptake in the lobulated heterogeneously enhancing para-rectal mass abutting the right levator ani muscle (SUVmax 10.9), with mild FDG uptake in the mesorectal lymph node (SUVmax 3.6), suspicious for nodal involvement, and no evidence of FDG avid distant metastasis (Figures 2 and 3).

Endoscopic trans-rectal core biopsies were performed. Histopathology showed a multilayered transitional and squamoid epithelium with underlying fibrous tissue and a few incomplete bundles of smooth muscle. Rectal glands

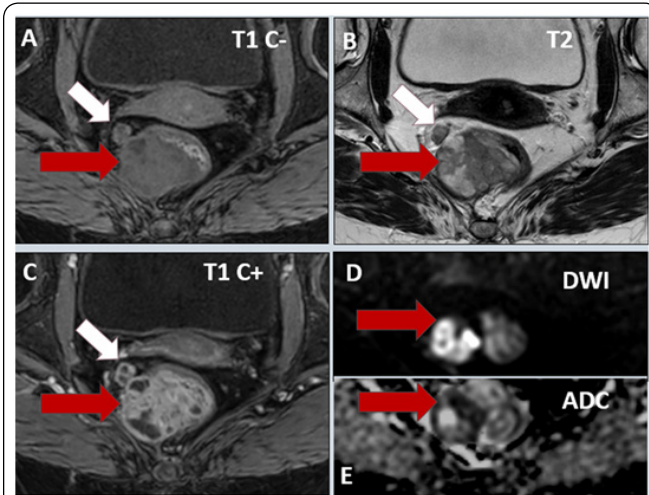


Figure 1: MRI showing multiple internal septations with homogeneous enhancement (A & C) and restricted diffusion (D & E) with cystic components [T1 hypointense (A), T2 hyperintense (B), and non-enhancing (C)]. An adjacent suspicious enhancing lymph node was noted (white arrow). The rectum is displaced anteriorly.

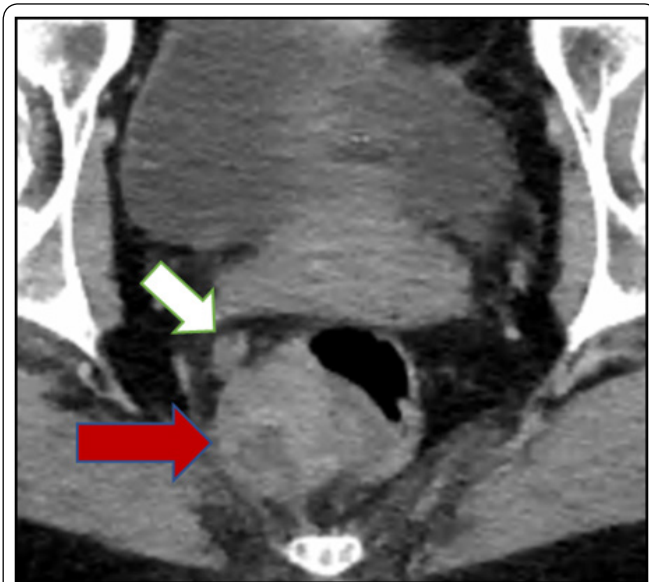


Figure 2: Contrast enhanced CT demonstrating a 4 cm lobulated para-rectal lesion (red arrow) with adjacent enhancing 8 mm lymph node (white arrow).

with goblet cells or dermal appendages were not seen. The epithelium demonstrated cytological atypia with an increase in mitoses. There were a few atypical cells infiltrating into the underlying stroma and muscle (Figure 4). A transitional cell / squamoid carcinoma, likely arising from a tailgut cyst was favoured.

Discussion

Tailgut cysts, traditionally known as retrorectal cystic hamartomas, are thought to originate from aberrant incomplete involution of tailgut remnants during embryogenesis [1, 2]. Often found incidentally, they are associated with a normal appearing rectal mucosa on lower endoscopy [3], with an

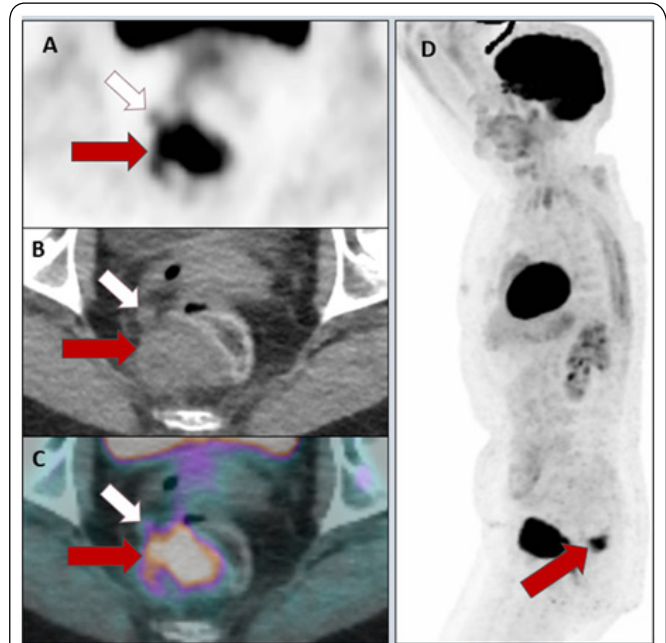


Figure 3: FDG PET showed the para-rectal mass was intensely FDG avid (SUVmax 10.9, red arrow) with mild uptake in an adjacent mesorectal lymph node (SUVmax 3.6, white arrow), suspicious for malignancy.

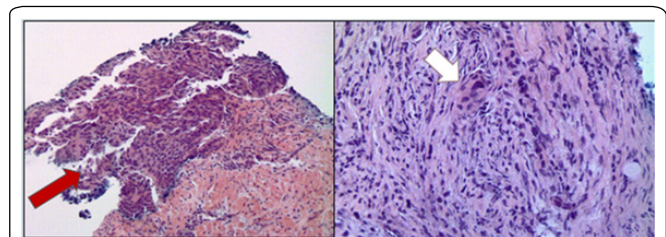


Figure 4: Histopathology showed atypical transitional cell and squamoid epithelium with an increase in mitoses (red arrow) and a focus of infiltration into underlying smooth muscle (white arrow).

increased frequency in middle-aged females [4].

Tailgut cysts are typically para-rectal lesions. Imaging appearances include a uni or multi-loculated, well-margined, thin-walled hypoattenuating cystic lesion on contrast-enhanced CT with possible calcification [1, 2]. There may be abnormalities in adjacent sacral bone suggestive of a developmental abnormality. MRI, considered the best modality for imaging these lesions to delineate its septations [4], typically demonstrates hypointense T1 and hyperintense T2 signal with low T2 intensity linear septations internally and no signal loss on fat saturated images [1, 4]. Intermediate signal intensity on T1 and T2 weighted images with loss of defined margins may indicate malignant degeneration although this may vary with protein content [1, 4]. Endoscopic ultrasound may confirm the presence of complex anechoic cystic components with internal echoes due to mucoid material [1]. These imaging findings are not specific for tailgut cysts, and differential diagnoses include dermoid/epidermoid cysts, duplication cysts, cystic sacrococcygeal teratoma, foreign body reactions, endometrioma and neoplasms including stromal, neurogenic and anorectal tumours [2].

The macroscopic appearance of a tailgut cyst is of a well-circumscribed lesion, which is typically multiloculated, ranging in size from 2 cm - 12 cm and containing clear serous or translucent thick mucoid fluid [5]. Microscopically, tailgut cysts are lined by any type of adult/foetal gastrointestinal tract epithelium with the most common being stratified squamous epithelium but may also contain transitional, cuboidal, mucinous or ciliated columnar epithelium. They have an underlying fibroconnective tissue stroma with disorganised smooth muscle bundles [5].

The main histological differentials for this lesion include a rectal duplication cyst, epidermoid/dermoid cyst and cystic sacrococcygeal teratoma. Duplication cysts and tailgut cysts differ on histopathology. Duplication cysts contain a well-formed double layer of smooth muscle associated with a neural plexus and are usually unilocular, found in the prerectal location in continuity with rectal mucosa and lined by gastrointestinal tract epithelium (with villi, crypts and glands) [2]. Although a limited tissue sample obtained, the diagnosis of a tailgut cyst was favoured over a duplication cyst with the presence of a variety of epithelia and disorganized smooth muscle fibres within the cyst wall and corroborative endoscopic and radiological data. Epidermoid / dermoid cysts are usually lined by squamous epithelium with no glandular or transitional epithelium and may contain dermal appendages [2]. Cystic sacrococcygeal teratomas contain derivatives of two or more embryonic germ layers (ectoderm e.g. skin, endoderm e.g. glands and mesoderm e.g. muscle) [2] and although this diagnosis could not be definitively excluded, it was thought to be less likely in this case.

In a systematic review of 196 cases of tailgut cysts, malignant degeneration was reported in up to 26% of cases [6]. When a tailgut cyst is detected, complete surgical resection (including a portion of normal bowel) is recommended to exclude underlying malignancy [7]. Whether or not to biopsy is controversial due to concern about infection and seeding of malignant cells [8].

In this case, the FDG-PET scan raised suspicion for malignancy which was confirmed on biopsy. Treatment options considered included neoadjuvant chemoradiotherapy followed by surgery, or surgery alone. Following a multidisciplinary discussion with colorectal surgery, radiation and medical oncologists, a decision was made to treat the lesion as a squamous cell carcinoma. Definitive concurrent chemoradiotherapy was recommended with surgery reserved for incomplete response. Concurrent chemotherapy (IV mitomycin and oral capecitabine) and radiotherapy (55Gy/30) was given, complicated by acute toxicities of radiation proctitis, mild radiation cystitis, radiation dermatitis and coccygeal

discomfort, all of which resolved. Repeat FDG-PET scan at one and six months following the completion of treatment demonstrated ongoing complete metabolic and anatomic response.

Conclusion

Tailgut cysts are rare embryologic remnants with potential for malignant transformation. Although a wide differential exists for complex cystic lesions connected to the alimentary tract, careful radiological evaluation and corroborative histopathology assist to confirm the diagnosis. Resection is generally recommended for tailgut cysts, unless there is evidence of locally advanced disease. To our knowledge, this is the first reported case of malignant (squamoid / transitional cell carcinoma) transformation of a likely tailgut cyst diagnosed on FDG-PET and treated curatively with definitive chemoradiotherapy.

Disclosure statement

The authors declare no conflict of interest or acknowledgements.

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