Case Report

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Pathology Section

Malignant Transformation in Tailgut Cyst Presenting as Retrorectal Adenosquamous Carcinoma: A Case Report

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ABSTRACT

Tailgut cysts are rare congenital retrorectal lesions that can rarely undergo malignant transformation. Occurring predominantly in middle-aged females, these lesions are mostly asymptomatic, with rare symptoms arising mostly in association with malignant transformation. The mainstay of therapy is surgical excision with adequate clear margins. Postoperative radiotherapy is indicated in cases with involved or close margins postoperatively, high histologic grade of malignancy, presence of perineural or lymphovascular invasion or high proliferative index of tumour, in order to prevent local site recurrence. In this study, we present a case of adenosquamous carcinoma arising from a retrorectal tailgut cyst. The patient was treated with surgical excision followed by adjuvant chemoradiotherapy. Patient is currently doing well and is on routine follow-up as per institutional protocol.

Keywords: Adenocarcinoma, Immunohistochemistry, Retrorectal hamartoma, Squamous cell carcinoma

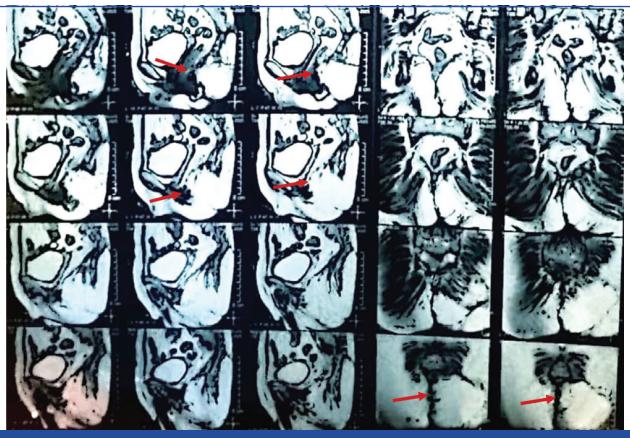
CASE REPORT

A 63-year-old female presented to the department of surgical oncology of our institute with a complaint of swelling in the retrorectal space for a period of six months. Patient gave history of a small papule with occasional discharge in that area from birth, which progressively increased in size to reach the present size for the last 6 months. There was no significant past surgical or medical history or any co-morbidity in the patient. Any significant family history was also not reported by the patient or her kin.

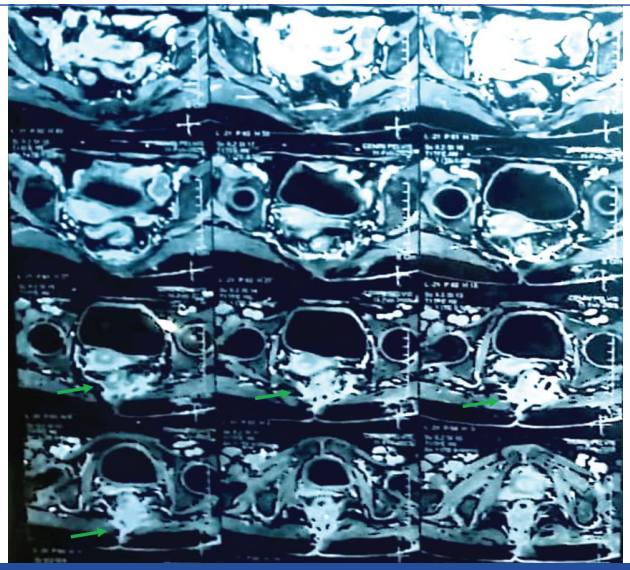
Local examination showed a (5×5) cm-sized ulceroproliferative lesion in the intergluteal cleft 4 cm above the anal opening, partly fixed to the underlying fascia. The lesion bled on touch. Systemic examination showed no signs of distant spread of disease. The tumour was given cTNM stage of cT3N0.

Contrast-enhanced magnetic resonance imaging of the pelvis showed a (6.3×5.6×5.5) cm space-occupying lesion in the coccygeal area, which showed a low signal intensity on T1-weighted images and a heterogeneously enhanced signal intensity on T2-weighted images [Table/Fig-1]. Post-Gadolinium scan showed a heterogeneous enhancement in the lesion [Table/Fig-2]. The lesion was seen to involve the levator plate with erosion of the tip of the coccyx. No significant intra-articular, neurovascular, or nodal involvement was identified radiologically. Bowel loops and rectum were unremarkable.

Histopathological examination of an incisional biopsy from the lesion revealed dermal infiltration by malignant glands with focal epidermal involvement and ulceration. A diagnosis of cutaneous involvement by adenocarcinoma was reached. Possible differentials considered were rectal adenocarcinoma with retrorectal skin infiltration or somatic type malignancy arising from sacrococcygeal teratoma. In view of the possible differentials, an urgent colonoscopy and a Positron Emission Tomography (PET) scan were advised for the patient. Any benign cystic conditions commonly seen in the anorectal area, such as anal anal gland cyst duplication cysts, were ruled out. The presence of a malignant tumour with a prominent adenocarcinoma component ruled out the possibility of chordoma, a tumour commonly arising at this site.

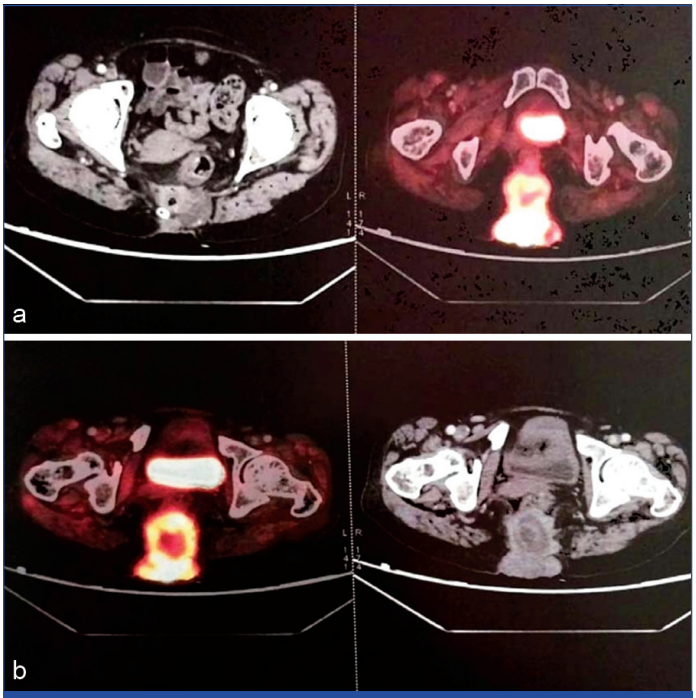


[Table/Fig-1]: Contrast-Enhanced Magnetic Resonance Imaging (CEMRI) pelvis showing a heterogeneously enhancing Space-Occupying Lesion (SOL) involving the coccygeal region with involvement of the levator plate and erosion of coccygeal bone (red arrows). The lesion shows low signal intensity on T1 and heterogeneous hyper-enhancement on T2 images.

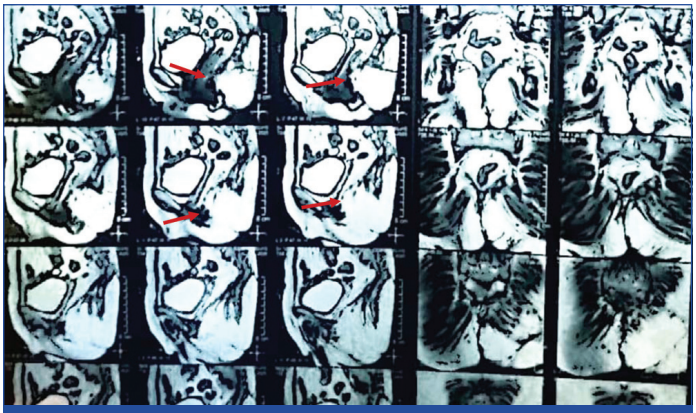


[Table/Fig-2]: Post-Gadolinium scan showed a heterogeneous enhancement in the lesion (green arrows).

Colonoscopy carried out in this patient was, however, unremarkable. Whole body Gallium 68 (68 Ga)–labeled Fibroblast Activation Protein Inhibitor (68Ga-FAPI) Positron Emission Tomography-Computed Tomography (PET-CT) was carried out. It showed a large, FAPI avid, lobulated, heterogenous soft-tissue mass in the sub-coccygeal region, involving the gluteal cleft, posterior pelvic wall and abutting the posterior wall of the rectum. The lesion showed central necrosis. There was associated cortical erosion of the lower end of the coccyx. The lesion measured 6.62x7.45x6.94 cm and showed a SUVmax of 19.64 [Table/Fig-3,4]. No other FAP expressing disease was identified in the rest of the body. FAPI PET-CT was preferred over Fluorodeoxyglucose (FDG) PET-CT due to its higher specificity and lower incidences of physiological uptake.



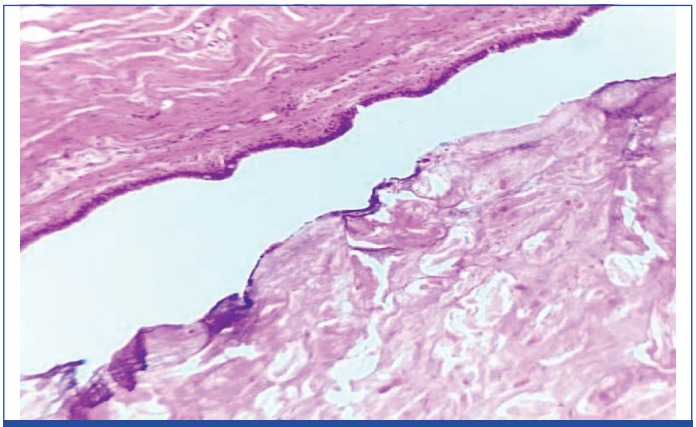
[Table/Fig-3]: a,b]: Whole body 68Ga-FAPI PET-CT was carried out. It showed a large, FAPI avid, lobulated heterogeneous soft-tissue mass in the sub-coccygeal region, involving the gluteal cleft, posterior pelvic wall and abutting the posterior wall of the rectum.



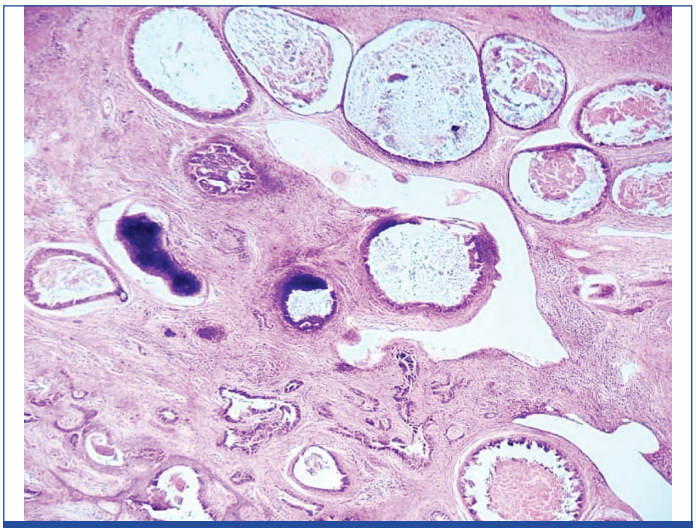
[Table/Fig-4]: Gross appearance of resection specimen of retrorectal tailgut cyst.

Macroscopic examination of the excision specimen showed a large proliferative tumour involving the skin, measuring 7 cm in maximum dimension [Table/Fig-4]. Cut surface of the tumour showed a solid cystic appearance with mucoid cystic content. The tumour was grossly seen to erode the coccygeal bone focally. All skin resection margins were free grossly; however, the deep soft-tissue margin of resection was abutted by the tumour.

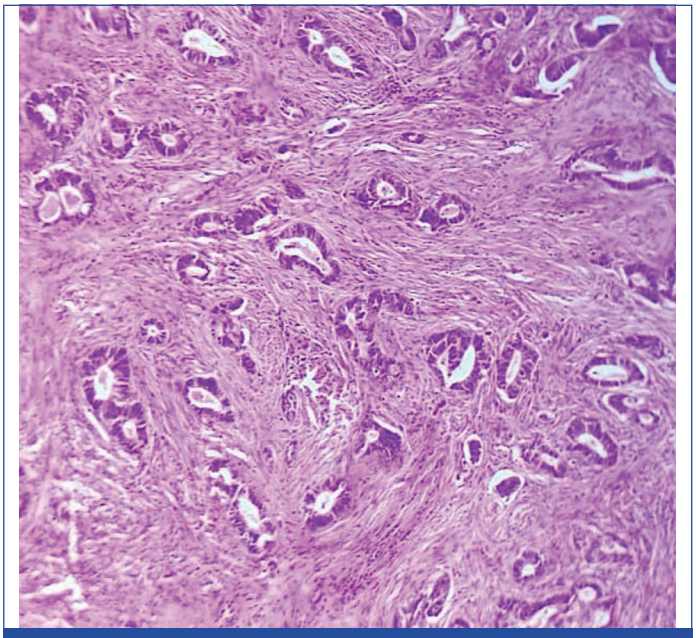
Microscopic examination of multiple sections showed a tumour mass lined by skin with smooth muscle proliferation and varying-sized cysts. The cysts were lined by cuboidal, columnar, ciliated, and squamous epithelium, containing keratinous debris in some of the cysts [Table/Fig-5,6]. The solid area showed an infiltrating tumour composed of dysplastic glands lined by pleomorphic columnar cells with hyperchromatic nuclei, as well as solid sheets of neoplastic epithelial cells with a squamoid morphology and keratin pearl formation [Table/Fig-7-9]. Both the adenocarcinoma and squamous cell carcinoma showed grade 2 differentiation, or were moderately differentiated. Skin infiltration and perineural invasion were identified [Table/Fig-10]; however, no lymphovascular invasion was seen. Superficial cortical erosion of the underlying coccygeal bone was seen [Table/Fig-11]. All skin resection margins were free, but the deep soft-tissue resection margin was close (<2 mm) to the tumour.



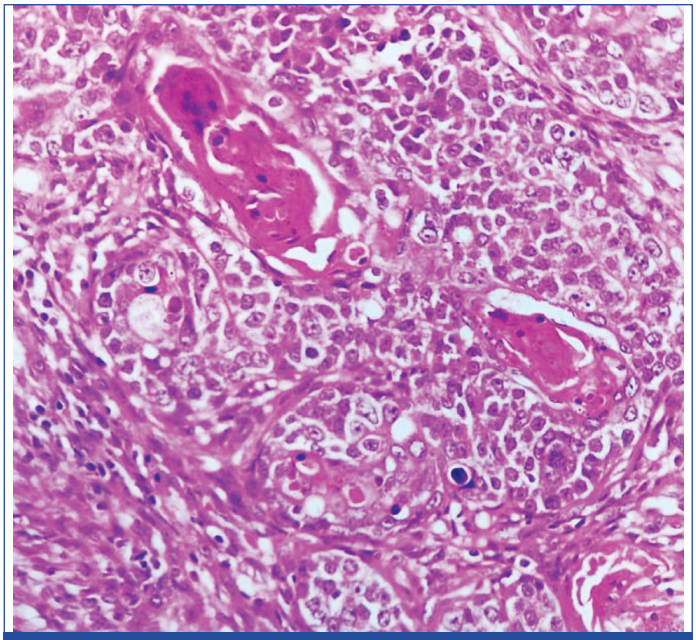
[Table/Fig-5]: Cyst lined by pseudostratified ciliated lining epithelium (10x), Haematoxylin and Eosin (H/E).



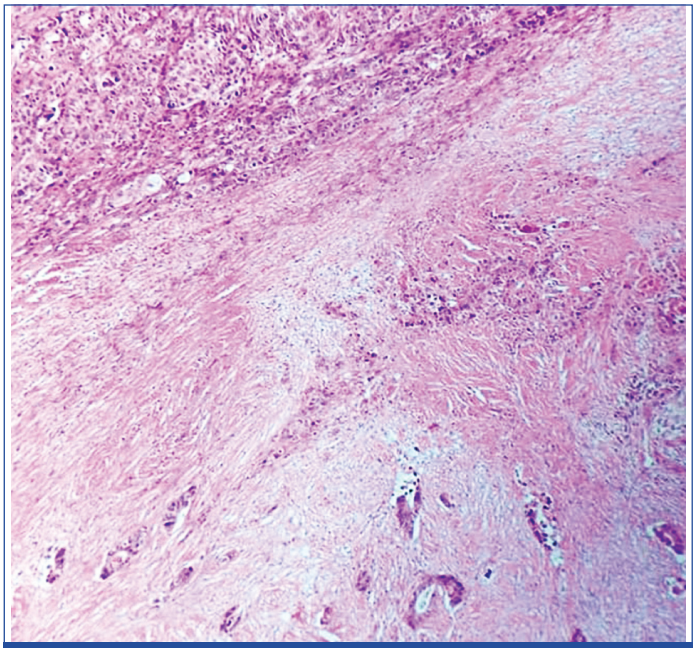
[Table/Fig-6]: Multiple cysts along with a focus of adenocarcinoma (10x), H/E.



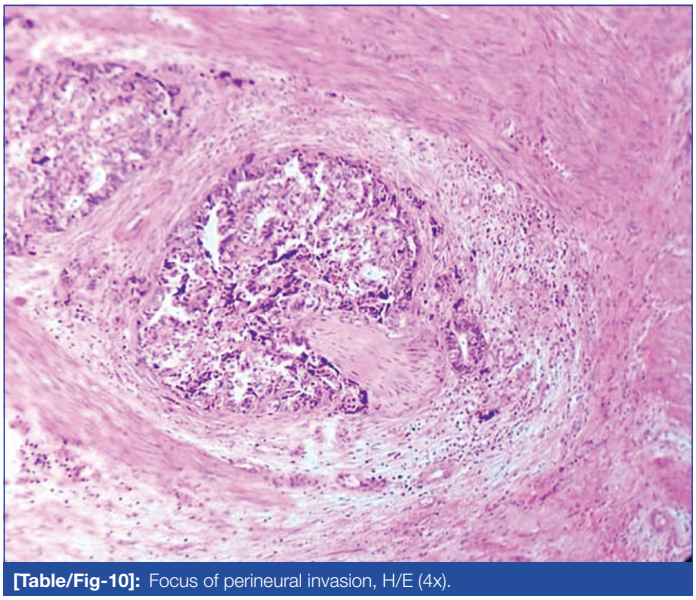
[Table/Fig-7]: Adenocarcinoma component (10x); H/E.



[Table/Fig-8]: Squamous cell carcinoma component (40x), H/E.

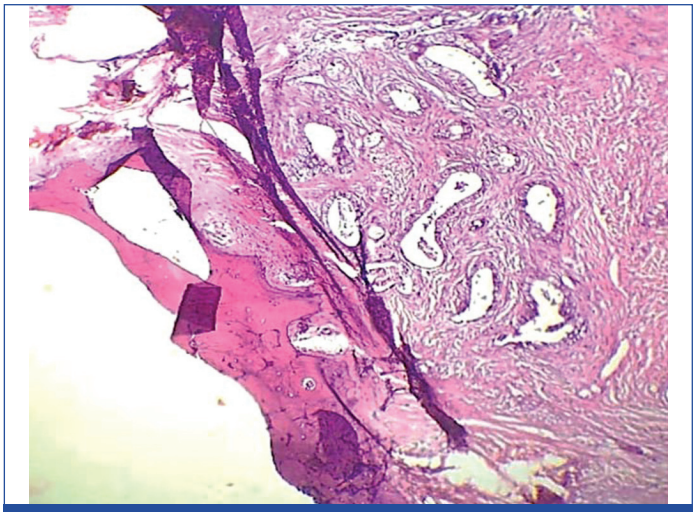


[Table/Fig-9]: Co-existent adenocarcinoma and squamous cell carcinomas (4x), H/E.

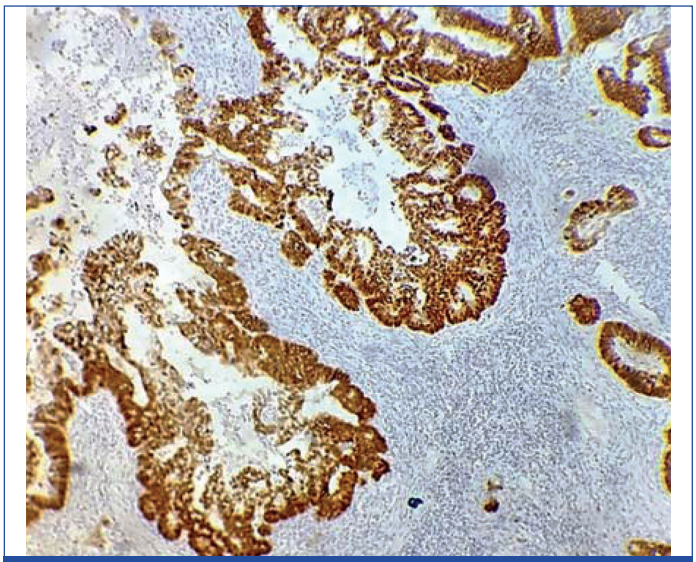


[Table/Fig-10]: Focus of perineural invasion, H/E (4x).

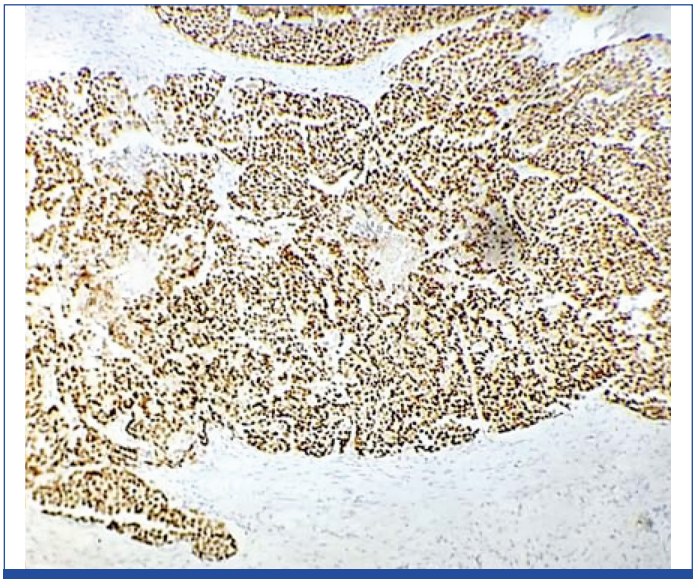
Immunohistochemistry showed strong diffuse positivity with CDX2 and p63 in the adenocarcinoma and squamous cell carcinoma components, respectively [Table/Fig-12,13], but was negative for Synaptophysin and CD56 [Table/Fig-14,15]. Positivity for CDX2 confirmed the adenocarcinoma component and P63 positivity confirmed the squamous cell carcinoma component. CD56 and Synaptophysin negativity ruled out a neuroendocrine tumour. Combined histomorphology and immunohistochemistry findings were consistent with an adenosquamous carcinoma arising in a background of tailgut cyst.



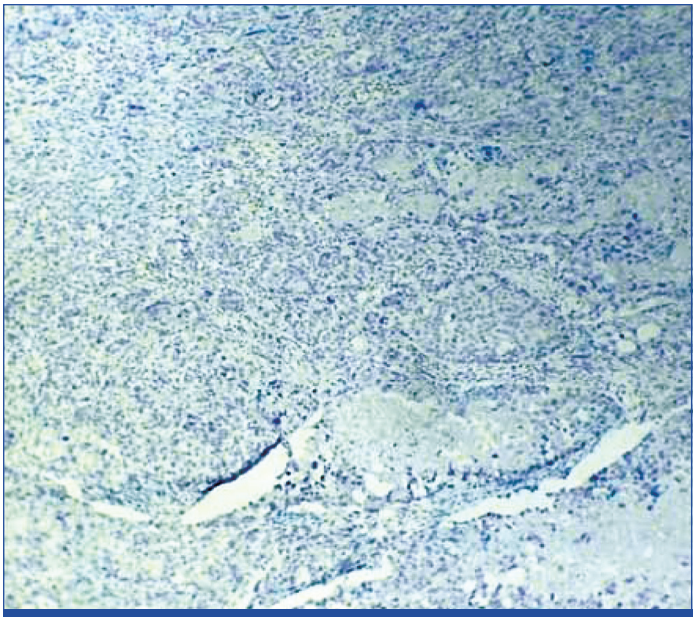
[Table/Fig-11]: Underlying bone invasion by malignant glands, H/E (4x).



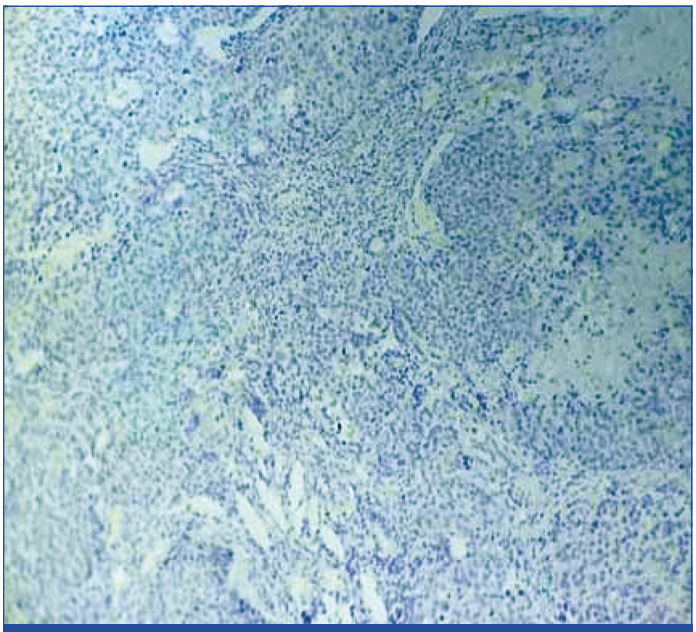
[Table/Fig-12]: CDX2 positivity in the malignant glands of the adenocarcinoma component, Immunohistochemistry (IHC).



[Table/Fig-13]: p63 positivity in the squamous cell carcinoma component, IHC.



[Table/Fig-14]: Negative for Synaptophysin, IHC.



[Table/Fig-15]: Negative for CD56, IHC (4x).

Following surgery, the patient was posted for adjuvant concurrent chemoradiotherapy. Patient received external beam radiotherapy to the postoperative bed with appropriate margins, along with concurrent chemotherapy with Capecitabine 825 mg/m2 twice daily on the days of radiotherapy. A total dose of 50.4 Gy was delivered in 28# at the rate of 1.8 Gy/#. Following adjuvant therapy, the patient was asked for follow-up, as per institutional protocol. At present, the patient is apparently doing well with no evidence of recurrence.

DISCUSSION

Tailgut cysts, also known as retro-rectal cystic hamartoma or epidermoid cysts, are cystic lesions developing in the presacral area and the retro-rectal space. They arise from remnants of embryonic hindgut, as a result of developmental abnormalities in the embryonic pathways [1]. Tailgut cysts are very rare, with an incidence of 1 per 40,000-63,000, most frequently affecting middle-aged females [2].

Tailgut cysts are polycystic lesions that always occur in the presacral space, bound by presacral fascia (Waldeyer fascia) posteriorly, fascia propria of rectum and mesorectum anteriorly, peritoneal reflection superiorly, levator ani muscles inferiorly, iliac vessels and ureters on the lateral side [3]. Histologically, these are multiloculated, lined by squamous, mucinous, simple, pseudostratified ciliated and transitional epithelium with occasional bundles of smooth muscles, meningeal tissue, thyroid tissue and glomus body [4].

The most common differentials of cystic lesions in this area include other developmental cysts like epidermoid, dermoid, duplication and non-developmental cysts like anal gland cysts and parasitic cysts. Neoplastic lesions like schwannoma, gastrointestinal stromal tumour and aneurysmal bone cysts have also been reported in this site [5]. Tailgut cysts should be differentiated from other lesions that are commonly seen in the retrorectal areas, such as teratoma, epidermal cysts, anal gland cysts and chordoma [6].

Clinically, tailgut cysts are mostly asymptomatic, with symptomatic cases presenting with compressive symptoms due to mass effect, such as constipation, rectal filling, dyschezia, infertility, dysuria and abdominal pain. Recurrent urinary tract infections, abscess and fistula formation have also been reported [7,8]. The most important features indicating a malignant transformation include cysts with symptoms, calcification, blurring of the cyst boundaries and encroachment of adjacent structures on imaging [9]. A histopathological review of 1708 cases of retrorectal lesions reported a 30% incidence of malignancy [10]. Another smaller case series of four cases reported an incidence of malignancy as high as 50% in tailgut cysts [11]. A recent analysis; however, reported a 9% incidence of neoplastic lesions in consecutive patients. The most common forms of malignancy arising from tailgut cysts include adenocarcinoma and neuroendocrine tumour. Others include carcinoid tumours, squamous cell carcinoma, endometrioid carcinoma, adenosquamous carcinoma, transitional cell carcinoma and rarely sarcoma [12-15]. Hormones like ghrelin and oestrogen might be important in the malignant transformation of tailgut cyst, possibly explaining the higher incidence among females. However, the exact pathogenesis is still unknown [16].

In our study, we reported a case of adenosquamous carcinoma arising from a tailgut cyst. This was reported once in literature [17]. Positivity for CDX2 confirmed the adenocarcinoma component and P63 positivity confirmed the squamous cell carcinoma component. CD56 and synaptophysin negativity ruled out a neuroendocrine component. The differentials considered in the present study, in view of the biopsy diagnosis of adenocarcinoma involving skin, were rectal adenocarcinoma extending to skin or somatic type malignancy arising from sacrococcygeal teratoma. Chordoma was not considered as a differential due to the absence of the classical histopathological findings.

CT scan and Magnetic Resonance Imaging (MRI) are the radiological evaluations of choice for diagnosis. MRI usually shows low signal intensity on T1-weighted images and high signal intensity on T2-weighted images [18]. Suspicious features of malignant transformation on MRI and CT include irregular mass contours, contrast enhancement within cysts, nodular wall thickening, calcification and infiltration of adjacent structures as seen in our case in the form of coccygeal bone erosion [9,19].

Complete surgical excision is the treatment of choice for all cases of tailgut cysts. Additionally, chemotherapy and radiotherapy can be provided if malignancy is detected on histopathological examination [20]. Indications for postoperative radiation also include elevated Ki67, presence of perineural invasion and close surgical margins to reduce the risk of local site recurrence [6]. Recurrence rates have been sparsely reported in literature, with a reported range of 0%-16% in literature [21]. Important prognostic factors include the time of diagnosis, the radical approach of the surgical procedure and the histopathological features of the resection specimen [22].

CONCLUSION(S)

Tailgut cysts presenting with malignant transformation can pose a significant diagnostic challenge due to their rare occurrence. Hence, in any case of retro-rectal cyst, where tailgut cyst can be

one of the differentials, surgical excision of the cyst along with adequate margins of resection should always be considered, owing to the possibility of malignant transformation. Complete surgical excision is the gold standard of treatment, along with postoperative radiotherapy in cases of close surgical margins, high-grade malignancy, increased proliferation rate or Ki67 index and presence of neurovascular invasion, to reduce the rates of recurrence.

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