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Case Report

Tail gut cyst adenocarcinoma: A rare entity diagnosed on cytology

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ABSTRACT

Tailgut cyst is a rare benign cystic lesion believed to originate from small portions of the embryonic tailgut, which normally disappear in early foetal development. It is often misdiagnosed due to the general unawareness with this entity and also because of its clinical presentation similar to other common diseases such as perianal fistulas or abscesses.

We take this opportunity to report a case of tailgut cyst in a 33-year-old Indian male. The patient had presented with painful swelling in sacral region, which had been gradually increasing in size. The case was diagnosed by fine needle aspiration cytology (FNAC) as adenocarcinoma arising in tailgut cyst and later confirmed by histopathology.

Tailgut cysts are congenital cystic hamartomatous lesions, which occur very rarely. They are generally benign, but malignant change or transformation in it has also been reported, which is even rarer in occurrence. FNAC is an effective diagnostic tool, which if applied appropriately can be used for the early detection of these lesions and also malignant transformation of tailgut cyst.

1. Introduction

The retro rectal cystic hamartoma or tailgut cyst is a rare congenital lesion derived from vestigial elements of the embryonic hindgut. Developmental cysts, which account for 60% of all congenital presacral tumors, can arise from any embryonic layer. Based on morphology, developmental cysts are classified into dermoid cysts, epidermoid cysts, tailgut cysts (TGCs) and cystic teratomas. [1,2]

Primary adenocarcinomas of the retro rectal or presacral space are uncommon and usually arise from the remnants of the embryological postnatal gut which contain mucous secreting epithelium. Clinical diagnosis is usually delayed due to non-specific symptoms and histological diagnosis can be obtained only after a biopsy or surgery. In such situations FNAC is much of diagnostic help along with radiological findings. These cysts have potential for infection, perianal fistulas and most importantly malignant change; hence an early complete surgical resection is the treatment of choice. [3–5]

2. Case Report

A 33-year-old male presented with sacrococcygeal mass since childhood, increasing slowly, which became painful since last 1 year. Pain was dull aching more on sitting. Local examination revealed sacrococcygeal fluctuant, non-tender oval mass measuring 7x5x1 cm, having regular margin with extension between the gluteal cleft. CT scan revealed a large complex, solid cystic mass measuring 8x7x6.5 cm in the retro-rectal and pre-sacrococcygeal area extending outward in left medial gluteal region. Three differential diagnoses of sacrococcygeal teratoma, tail gut cyst and remote possibility of soft tissue sarcoma were offered by radiologist.

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FNAC done twice including once under USG guidance. At first 20ml of dirty brownish fluid aspirated and the diagnosis of tail-gut cyst was given after advising USG guided FNAC for solid component. Repeat FNAC under USG guidance from solid component showed sheets and clusters of round to polygonal cells with moderate amount of cytoplasm, nucleomegaly, prominent nucleoli, hyperchromasia, all features indicated malignant cells. Also seen were benign ciliated columnar epithelium of gut mucosa type with tumor cell necrosis, chronic inflammatory infiltrate and foamy macrophages in the background. The diagnosis of malignant transformation to adenocarcinoma in tail gut cyst was given on FNAC. Patient had undergone resection surgery for the tumor and histopathology confirmed the diagnosis of invasive adenocarcinoma arising in a tail gut cyst.

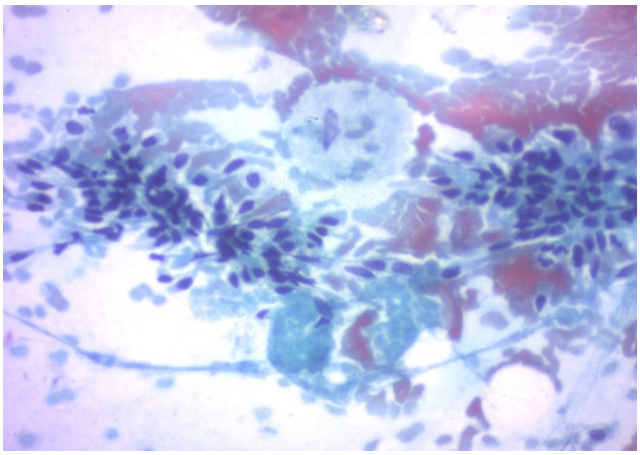

Figure 1: FNAC from cyst showing ciliated columnar epithelium (Pap10X)

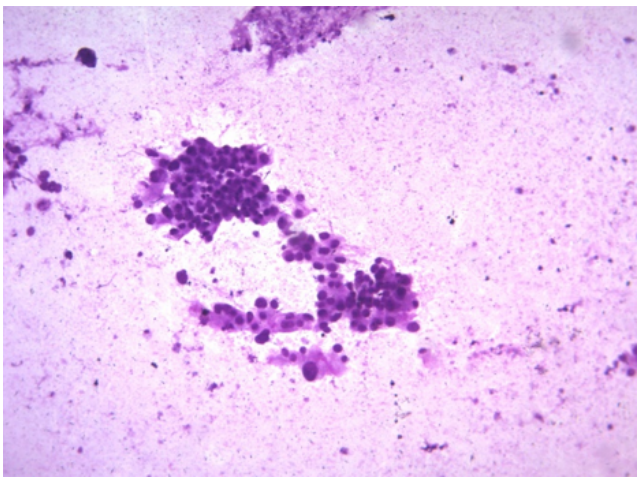

Figure 2: FNAC showing adenocarcinoma cells with anisonucleosis and hyperchromasia (MGG10X)

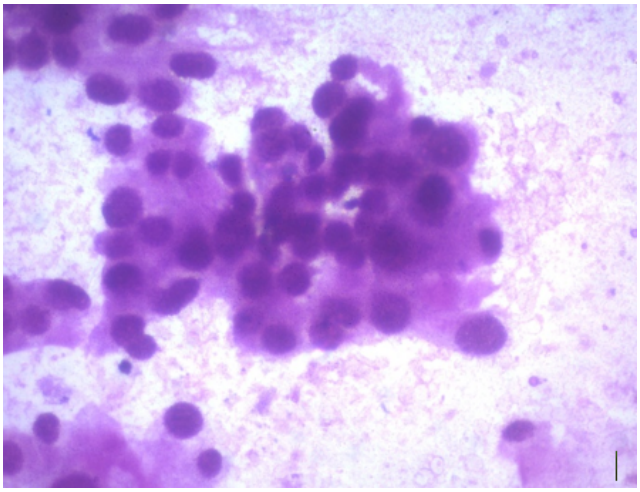

Figure 3: FNAC showing adenocarcinoma cells with anisonucleosis and hyperchromasia (MGG 40X)

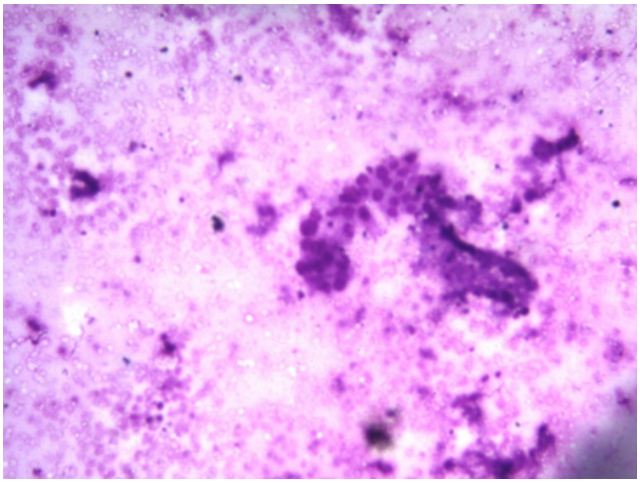

Figure 4: FNAC showing necrosis of tumor cells in the background (MGG10X)

3. Discussion

The retro rectal cystic hamartoma or tailgut cysts (TGC) are rare congenital lesions derived from the elements of the embryonic hindgut. Embryo possesses a true tail caudal to the site of subsequent formation of anus. The primitive hindgut extends into this tail, which usually completely regresses. However, sometimes tailgut remnants in the retro rectal space persists giving rise to tail gut cyst.[1] Congenital lesions are by far the most common tumor type in this region, accounting for 55-70% of all presacral tumors. These tumors are present from birth because of the growth of embryological remnants, and they include cystic (developmental cysts, rectal duplication cysts, and anterior meningoceles) and solid (chordomas, teratomas, and adrenal rest tumors) lesions.[1,2,5,6]

TGCs, are rare benign cystic lesions of the presacral space,usually presents as asymptomatic mass but can also present with dull aching pain, rectal bleeding, pain on defecation, urinary problems or lower back pain.[2] It may get secondarily infected, and often diagnosed as anorectal fistula, pilonidal cyst, or recurrent retro rectal abscess.[3] Major differential diagnosis like dermoid cyst, epidermoid cyst, rectal duplication cysts, and teratoma are excluded by considering the structure and lining epithelium of the cysts.[4,5] Presence of local invasion is indicative of malignancy. Review of literature shows that malignant change within a retro rectal cystic hamartoma had been documented in only 17 cases.[3–6] Owing to the rarity of the condition and its nonspecific presentation, diagnosis and thereby treatment is frequently delayed.

Tailgut cysts are usually multilocular and are lined by columnar or cuboidal epithelium. Ciliated, squamous, and transitional epithelium has also been noted. Acute or chronic inflammation is seen in around 50% of cases. A definite muscular and serosal coat is not present, although scattered bundles of smooth muscle fibers are seen in the cyst wall. Most cases are diagnosed in adulthood and 75–90% have been described in females.[1,2,7] Teratomas and dermoid cysts have also been seen in this area and usually comprise components of all the three germ layers. Although tailgut cysts also contain structures of all three germ layers (epithelium, fibrous tissues blood vessels, smooth muscle), ciliated epithelium also occurs in the embryo's GI tract and is not exclusively of respiratory origin.[2,7]

According to Hjermsad et al, the following histological criteria have been established for the diagnosis of TGC: (1) the epithelial lining of the luminal surfaces of the cysts must contain transitional and/or glandular-type (columnar) epithelium, with or without stratified squamous components; and (2) the underlying stroma must be composed of fibrous connective tissue containing scattered, discontinuous bundles of smooth muscle fibers, without a well-defined muscular layer containing a myenteric plexus and serosa.[8] Although radiological tools such as transrectal ultrasonography (TRUS), computed tomography (CT)/ magnetic resonance imaging (MRI) provide sufficient information for the differential diagnosis of most presacral TGCs, histopathological examination is always required for a definite and accurate diagnosis.[9]

Many patients with tailgut cysts probably remain asymptomatic till adulthood. Tailgut cysts, however, may grow to considerable size and present with gynecological and obstructive urinary or rectal symptoms, chronic abscesses and fistulas, and in extremely rare cases with malignant transformation, as in our case. It is believed that dysplasia-carcinoma sequence, which is established in colon also exist in tailgut cyst.[6,7] The first case of a tailgut cyst with malignant epithelial transformation was described by Ballantine in 1932 and since then about 17 cases of this rare phenomenon have been documented; the cases described were all adenocarcinomas or carcinoid tumours.[8,9] Adenocarcinoma arising in TGC has been associated with an elevated serum CEA and/or serum CA 19-9 levels. However, CEA elevation as such is not specific enough to permit a diagnosis of TGC adenocarcinoma. CEA levels may be used as a measure to assess the tumour's response to treatment or development of local recurrence and distant metastasis. Hence these lesions should be excised early and completely to reduce the risk of malignant change and other complications.[8,9]

The outcome of tail gut cysts (TGC) associated malignancies has varied from case to case. The factors that determine the prognosis have been thought to be the age at diagnosis, completeness of resection, tumour histology and grade; with neuroendocrine (or carcinoid) tumors having better prognosis than adenocarcinomas. Many cases of adenocarcinoma arising from TGC have reported the poor prognosis due to local recurrence and metastasis.[7,9]

4. Conclusion

Primary adenocarcinomas of the presacral /retro rectal space are uncommon and usually arise from cystic lesions developing from remnants of the embryological postanal gut [tail gut cysts (TGC)] containing mucous-secreting epithelium. Clinical diagnosis is usually delayed by non-specific symptoms and FNAC is an effective diagnostic tool, which if applied appropriately can be used for the early detection of these rarer lesions such as tailgut cyst. Histopathological diagnosis is confirmatory and definitive but usually obtained after surgery only. The potential for infection, perianal fistulas and perhaps most importantly, malignant change emphasizes that an early complete surgical resection is the treatment of choice.

5. Author's Contribution

Both authors contributed equally in conceptualization, preparing, editing and formatting of the manuscript

6. Conflict of Interest

The authors declare no conflict of interest.

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None.

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