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Case Report

Basaloid squamous cell carcinoma of the anus: a case report

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ABSTRACT

Anal cancer, which affects the anus, anal canal, or anorectum, represents ~2.2% of all gastrointestinal malignancies. Squamous cell carcinoma (SCC) is the most common type of anal cancer. Basaloid SCC is a rare and aggressive variant of the SCC, which is mainly seen in the upper aero digestive tract and the anal canal. These carcinomas are characterized by marked histological undifferentiation and a heightened proliferation rate. Macroscopically, appear as a perianal ulcer or a mass with raised edges and ulceration, often misdiagnosed as hemorrhoidal disease. Despite its rarity, accurate diagnosis is crucial. In the literature, there are only few case reports with such aggressive cancer having good outcomes with chemoradiation therapy. Owing to its infrequency, we present a case of basaloid SCC of the anus in a female patient with poor outcomes, alongside a comprehensive literature review.

Keywords: Basaloid squamous cell carcinoma, Anal carcinoma, Anus, Histologic subtype

INTRODUCTION

Anal cancer is a type of cancer that forms in the tissues of the anus. Although considered to be a rare cancer, the incidence rate of invasive anal carcinoma is increasing worldwide. Most anal cancers are related to human papillomavirus (HPV) infection.[1] The differential diagnoses of anorectal cancers include SCCs, adenocarcinomas, and neuroendocrine carcinomas. The most common histologic type is SCC. Basaloid SCC (BSCC) is a rare variant of SCC occurring in the anorectal region. It has a predilection for the upper aerodigestive tract.[2]

BSCC of the anus usually arises from the transitional epithelium of the dentate line. Clinically often mimics a rectal adenocarcinoma, present as a distal rectal or perianal mass, although the etiopathogenesis and histopathology of the two are distinct.[3]

Basaloid squamous carcinoma tends to have an aggressive clinical course with frequent local recurrences with regional and distant metastases. It has been reported that the common sites of metastatic spread are lymph nodes, liver and lungs.[4,5] Most BSCCs are usually diagnosed at advanced clinical stages with unfavorable prognosis.

Staging recommendations for anal cancers are based on tumour size, lymph nodes involvement and tumour spreads. Clinical examination including DRE and inguinal lymph nodes evaluation should be carried out for the assessment of tumour extent.[4] Contrast-enhanced computed tomography (CT) scanning of the thorax, abdomen and pelvis is a requirement in all patients to assess potential metastatic disease sites at diagnosis and follow-up.

The treatment landscape for basaloid SCC of the anus is developing, with CRT remaining the cornerstone of therapy.[4] Salvage surgery is only feasible for locally residual or recurrent disease. Given the rarity of the disease, it is important matching patient preferences with ideal treatment options.

We present a case of a 43-year-old female that presented with hematochezia leading to a diagnosis of lower gastrointestinal bleeding. Colonoscopy was performed and showed an ulcerated and friable lesion with histopathological examination consistent with invasive basaloid SCC.

CASE REPORT

The patient is a 43-year-old female with chief complaint of intermittent hematochezia for two years. The patient also noticed recent weight loss. She has no relevant medical history, except meningitis in childhood and epilepsy. There was no family history of colorectal malignancy.

Digital rectal examination revealed a friable polypoid lesion in the anal canal. As part of the diagnostic evaluation, a colonoscopy with biopsy was performed, revealing an ulceroinfiltrative anorectal mass-like lesion. The histopathological analysis indicated a poorly differentiated SCC with basaloid features. Microscopic examination showed malignant nests of cells with peripheral nuclear palisading. Immunohistochemistry confirmed positivity for p63. The pelvic magnetic resonance imaging (MRI) revealed a 43 mm tumour in the anal canal extending to the low rectum and affecting the internal sphincter. Also showed multiple enlarged lymph nodes in the para-aortic, iliac, obturator, inguinal and perirectal area (Figures 1).

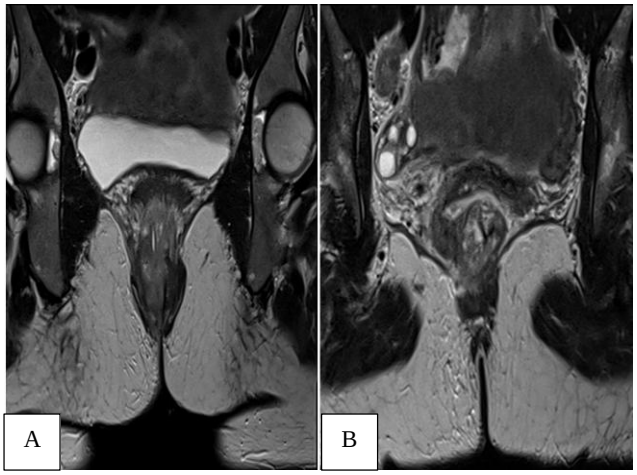

Figure 1 (A and B): T2 MRI coronal view of the mass shows invasion of the internal sphincter complex.

Computed tomography of the chest, abdomen, and pelvis demonstrated multiple masses compatible with metastasis at lungs, liver and bones; anorectal mural thickening with infiltrative changes in the adjacent mesorectal fat and multiple lymph nodes measuring 1 cm (Figure 2).

Subsequently, treatment was decided in a multidisciplinary team meeting. The patient was started palliative chemotherapy with disease progression and death.

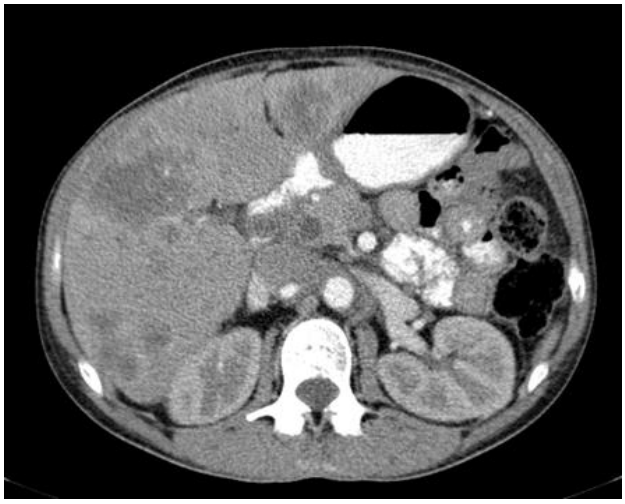

Figure 2: CT scan with intravenous contrast shows multiple liver metastasis.

DISCUSION

Anal carcinomas are uncommon and account for approximately 2.2% of gastrointestinal cancers. BSCC is a rare variant of SCC occurring in the anorectal region.[5] It is estimated that 0.2% of all anorectal neoplasms are basal cell carcinomas.[5,6]

BSCC was first reported by Pang et al, Basaloid tumour is a histologic subtype of SCC arising usually in the anal canal, characteristically from the endodermal-ectodermal junctional zone (cloacogenic zone) of anus.[7]

These carcinomas are characterized by significant histological undifferentiation and high grade proliferation.[8] Chetty et al reported that BSCC of the anal canal is characterized by hipercromatic basaloid cells and tumour nests exhibiting eosinophilic infiltration.[9] BSCC are often misdiagnosed because they have several histological patterns.[5,8-10] Immunohistochemistry can be performed for differential diagnosis.[6-10] Graham et al reported that the tumour cells were negative for chromogranin, synaptophysin, S100 protein, and KIT, and positive for the squamous marker CK5/6 and p63.[10] Our case presented the typical morphology of a basaloid carcinoma.

This disease tends to manifest in the sixth decade of life, and more prone to affect females with a prevalence female-to-male ratio of 2:1.[5,7,8,10,11] Although considered to be a rare cancer, the incidence rate of invasive anal carcinoma is increasing due to various risk factors such as HPV infection, which is identified in 90% of patients, autoimmune disorders, hematologic malignancies, immunosuppression related to HIV infection or transplantation and smoking.[12]

Clinical presentation frequently includes hematochezia and anal pain; however, diagnosis may be delayed because bleeding is attributed to hemorrhoids. May also

present with any combination of a mass, non-healing ulcer, itching, discharge, faecal incontinence, and fistulae. These aggressive tumours are often presented and diagnosed at an advanced stage, with 50% having nodal or distant metastasis, leading to complications such as bleeding and bowel obstruction.[4,13]

The primary aim of treatment is to achieve cure with locoregional control, preservation of anal function and the best possible QoL. A secondary aim is to manage symptoms related to the cancer and treatments late effects. A multidisciplinary approach is mandatory, involving radiation oncologists, medical oncologists, surgeons, radiologists and pathologists. The role of surgery as a salvage treatment is accepted.

Systemic therapy, paclitaxel plus carboplatin, is the usual approach for treatment of metastatic anal squamous cell cancer.[4] Immunotherapy using agents that target programmed cell death receptor 1 pathway is option for patients who have progressed on 1st line chemotherapy for metastatic SCC of anus. Further immunotherapy-based approaches are currently underway.[4]

Twenty percent of patients develop local failures following CRT, and salvage surgery is only feasible for a proportion of such patients.[4] The prognosis of all metastatic patients is poor with a 5-year relative survival rate of 30%.[6]

CONCLUSION

Anal cancer is an extremely rare clinical entity. SCC is the most common type of anal cancer. Basaloid SCC is a rare variant of the SCC with more aggressive behavior and poor prognosis. Most basaloid SCCs are extensively involved in metastasis to the lymph nodes, liver, and lung at diagnosis.

Identifying a BSCC represents a challenge due to its resemblance to other anal diseases and because it is a rare disease with limited cases presented worldwide. Therapeutic consensus has not been established since that are several therapies under investigation.

This case highlights the importance of a multidisciplinary approach in assessing the patient with a rare disease, presenting all viable options for treatment and electing the optimal treatment through shared decision making.

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