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Diagnosing Primary Squamous Cell Cancer of the Rectum in a Patient With HIV: A Case Report

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Introduction

Squamous cell cancer (SCC) of the rectum is a rare GI tract malignancy, with fewer than 80 reported cases and fewer than five case reports in patients with HIV.^{1,2} There is an increased risk of developing rectal SCC in patients with HIV.³ We present detailed history, pathology, and radiologic evidence of rectal SCC in a patient with HIV as a distinct entity separate from anal SCC.

Case Presentation

A 45-year-old African American male with a history of HIV, diabetes, and schizophrenia presented with rectal bleeding. He was diagnosed with HIV at age 23 years. He had a history of anal receptive intercourse with three male sexual partners. He never had AIDS-defining illness or a CD4 count less than 100. He was being treated with anti-retroviral medications with 100% adherence. A computed tomography scan of the abdomen revealed a 5.1 cm × 5.1 cm × 4.0 cm mass at the rectosigmoid junction. On colonoscopy, a 6-cm obstructive and bleeding mass approximately 7 cm from the anal verge was visualized. A positron emission tomography–computed tomography scan (Fig 1) revealed a large eccentric, hypermetabolic rectal mass without evidence of local or distant metastasis and no involvement of the anal canal.

Pathology revealed SCC of the rectum. It was diffusely p16-positive by immunohistochemistry (Fig 2). Tumor cells expressed

AE1/AE3, which is suggestive of squamous cell origin. It was CAM5.2-positive, which characteristically stains rectal SCC and adenocarcinoma but not anal squamous cell lesions.^{4,5} It was also positive for high molecular weight keratin and p63. This combination of positive markers confirms SCC related to human papillomavirus (HPV) infection with the rectum as the tissue of origin. This case report meets the three diagnostic requirements established by Williams et al⁶ for diagnosing histologically confirmed SCC of the rectum: the tumor was not a metastasis from a distant site, there was no squamous cell-lined fistulous tract, and the mass was not an extension of SCC of the anus.

Discussion

In the current literature, the pathogenesis of SCC of the rectum is not clear. As there is a known role of HPV in SCC of the anus, HPV could play a contributing role in the rectum, but reports are conflicting.^{3,5} In addition, immunosuppression is noted to be an important risk factor. There is a 20-fold increased risk for developing rectal SCC in HIV-infected patients with AIDS, especially in men who have sex with men.² This case report provides imaging and immunohistologic evidence to prove beyond doubt the diagnosis of primary SCC of the rectum. p16 stain is diffusely positive, which provides further support of the likelihood of HPV playing a strong role in SCC genesis. It is possibly associated with trauma

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Fig 1. Axial positron emission tomography–computed tomography image through the pelvis demonstrates a large eccentric, markedly hypermetabolic rectal mass. No regional adenopathy or distant metastasis was detected.

that leads to a high infection rate in patients with anal receptive sex. This case also supports the retrospective findings noted by Coghill et al³, as this patient fits the demographic profile of a positive history of male sexual partners and positive HIV status.

Currently, anal SCC is treated primarily with chemoradiation. Sturgeon et al¹ have reported excellent results in a series of 14 female patients with rectal SCC that was treated with chemoradiation. Following the tumor board recommendations, our patient was treated with chemoradiation (fluorouracil, mitomycin, and radiation therapy) for 6 weeks. The patient currently presents after therapy with resolution of symptoms and a significant decrease in the mass size. Although the future clinical course of our patient is unknown, should locoregional recurrence occur, the patient may be treated with salvage surgery; however, addition of high-dose intraoperative radiation therapy to salvage surgery was of little benefit in a series of patients with recurrent SCC of the anal canal and, therefore, may be of limited benefit in our case.⁷ **JOP**

Authors' Disclosures of Potential Conflicts of Interest

Disclosures provided by the authors are available with this article at jop.ascopubs.org.

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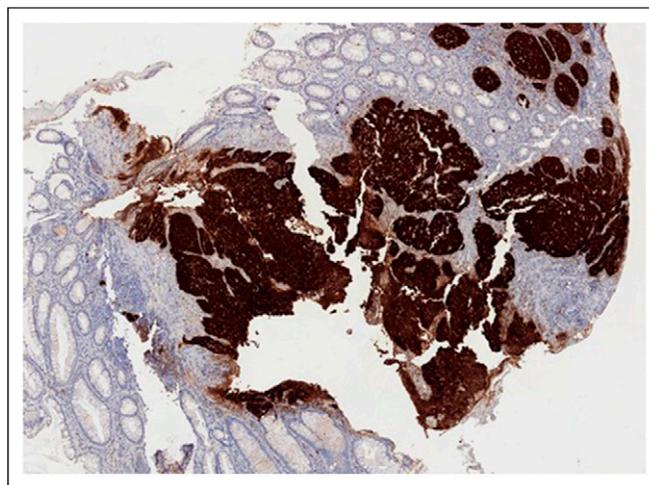


Fig 2. Diffusely p16-positive specimen consistent with squamous cell carcinoma likely related to human papillomavirus infection. Magnification, ×4.

Provision of study materials or patients: Irina Dobrostokaya

Collection and assembly of data: Pushpinderdeep S. Kahlon, Ashish Mishra, Todd R. Williams

Data analysis and interpretation: All authors

Manuscript writing: All authors

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AUTHORS' DISCLOSURES OF POTENTIAL CONFLICTS OF INTEREST**Diagnosing Primary Squamous Cell Cancer of the Rectum in a Patient With HIV: A Case Report**

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