

HPV 16 Positive Squamous Cell Rectal Carcinoma: A Rare Entity

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Introduction

Squamous cell carcinoma (SCC) of the rectum is an uncommon malignancy, accounting for approximately 0.10-0.25 per 100 colorectal neoplasms¹. HPV positive rectal cancer is an exceedingly rare subset of rectal malignancies as well.

Case Description

A 50-year-old male with past medical history of hypertension, Hepatitis C status post interferon-based treatment, active intravenous drug use, who presented with melena and abdominal pain. He reported that over the past two months he had noticed haematochezia and altered bowel habits. He has no family history of colorectal cancer or prior colonoscopy.

Physical exam: Well-developed and not in acute distress.

HENT: No scleral icterus. Conjunctiva normal.

Heart: RRR, no murmurs; **Lungs:** no rales or wheezes

Abdominal: Bowel sounds are normal. Abdomen is soft. There is no abdominal tenderness. There is no guarding or rebound.

Labs: Haemoglobin 12.4 with MCV of 86.3; Carcinoembryonic antigen 24.1.

Computed Tomography (CT) Abdomen/Pelvis: Large rectal mass filling almost the entire mid and lower meso-rectal envelope. The mass appeared to be centered along the rectal wall posteriorly with ventral displacement of the lumen. Enlarged superior rectal and right extra-meso-rectal lymph nodes.

MRI of the pelvis: Large mid-rectal tumour arising from the posterior wall of the rectum causing luminal narrowing and involvement of the meso-rectal fascia posteriorly with right extra-mesorectal enlarged lymph nodes.

CT Chest: Sub-pulmonary emphysema and sub-centimetre pulmonary nodules measuring up to 6 mm (3 mm right lower lobe subpleural nodule, 4 mm nodule along the minor fissure, and 6 mm right middle lobe nodule).

Pathology: Biopsy of the rectal mass confirmed invasive squamous cell carcinoma. Immunohistochemical stain was positive for p16, p40 and AE1/AE3. HPV 16 positive.

Diagnosis: Rectal Squamous Cell carcinoma stage T3N1M0

Treatment:

- Curative intent chemoradiation therapy with 5-Fluorouracil (5-FU) and Mitomycin C (MMC).

Results:

- Repeat MRI showed significant decrease in tumor size.
- Colorectal surgery reviewed and planned for salvage surgery.

Figure 1: Tumor in rectum prior to therapy (A). Tumor in rectum after chemoradiation (B)

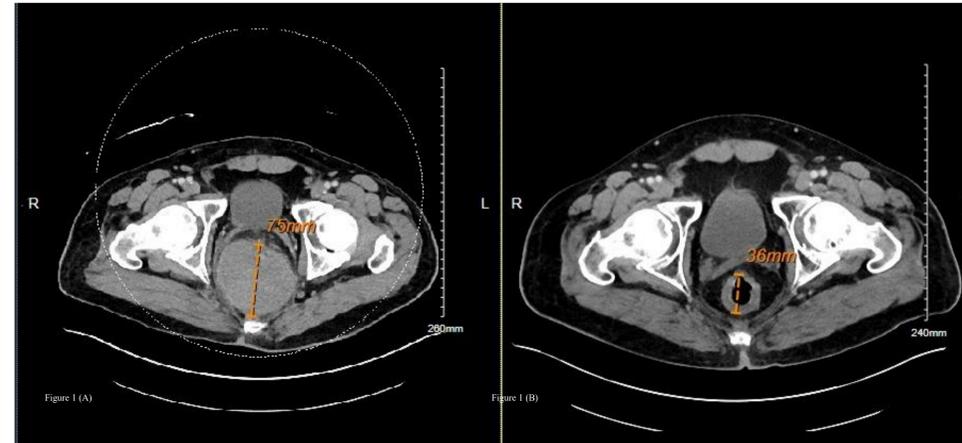
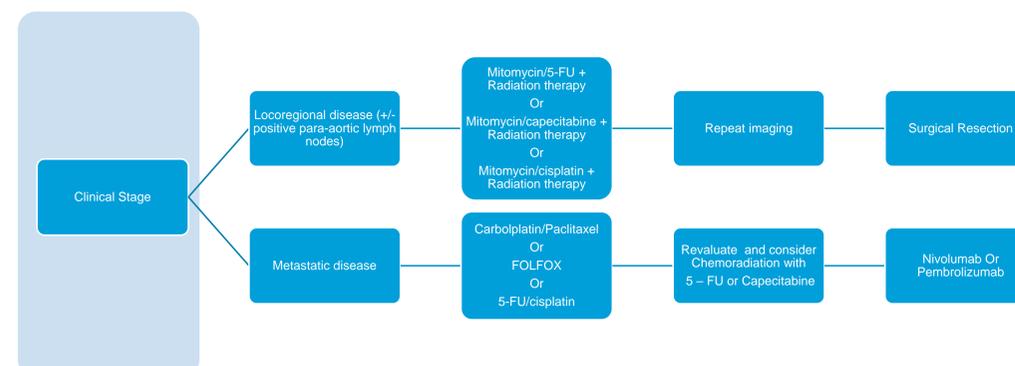


Figure 2: Colonoscopy notable for fungating partially obstructing mass with areas of ulceration in the distal rectum starting at the first valve of Houston. The mass was located between 5-15 cm from the anus.



Figure 3: Treatment of SCC of Rectum



Discussion

Epidemiology:

- Rectal cancer accounts for 35% of colorectal cancer¹.
- Subsequently, 90% of rectal cancers are adenocarcinomas, and the rest 10% consist of carcinomas, sarcomas, and lymphoid tumors¹.
- It affect individuals between the ages of 39 to 93 years old, with a mean age of 57 years².
- The disease tends to occur more frequently in women than in men¹.
- Patients often present with advanced disease, Stage II or III

Risk Factors:

- While no clear set of risk factors have been established, several associations have been observed such as IBD, HIV and HPV.^{2,3}

Common symptoms: Abdominal pain, weight loss, alteration in bowel habits, pain while defaecating and rectal bleeding.

Staging: Staging is similar to that of adenocarcinoma of the rectum and it is based on tumor size, location, number of lymph nodes and metastasis.

Prognosis:

- The overall 5-year survival for rectal SCC was found to be **48.9%** compared with **62.1%** for rectal adenocarcinoma.²
- Patients with HPV DNA or p16 positive anal SCC have significantly better outcome survival compared with HPV DNA or p16 negative.² However, clear association seen between HPV and rectal SCC has not been firmly established.

Treatment

- Due to the rarity of SCC of the rectum there is limited data on treatment of the disease.
- There is some evidence to support the use of 5-FU/MMC on Day 1- 4 and Day 29-32 with concurrent radiation therapy for treatment of rectal SCC, which is the evidence-based treatment used for anal SCC.^{3,4}
- Based on literature review, the consensus points towards treatment with curative intent chemoradiation and subsequent salvage surgery if there is not a complete response to initial treatment. Further large based prospective trials are needed to establish a standard of care however this is limited due to the rarity of the disease.^{3,4}

Reference

¹Caio et al. (2007). 'Squamous cell carcinoma of the rectum: A rare but curable tumor'. The American society of Colon and Rectal Surgeons.

²Astarras et al. (2021). 'Squamous rectal carcinoma: a rare malignancy, literature review and management recommendations'. ESMO Open.

³Rasheed et al. (2008). Chemoradiotherapy : an alternative to surgery for SCC of rectum'. The Association of Coloproctology of Great Britain and Ireland. Colorectal Disease.

⁴Guerra et al. (2016). 'Primary squamous cell carcinoma of the rectum: An update and implications for treatment'. World Journal of Gastrointestinal Surgery