Leiomyosarcoma of the rectum following pelvic irradiation: a difficult histological diagnosis

I Basu, P Lemonas

Princess Alexandra Hospital, Harlow, UK

ABSTRACT

Rectal leiomyosarcomas are very rare mesenchymal tumours. This is a case report of a rectal leiomyosarcoma diagnosed initially as a leiomyoma in a patient who had undergone pelvic radiotherapy several years previously. In addition to its pathological rarity, this case is of particular interest because it reinforces the association between pelvic irradiation and rectal leiomyosarcomas and it highlights the importance of treating suspicious cases aggressively in spite of favourable preoperative radiological and histological assessment.

KEYWORDS

Leiomyosarcoma – Rectum – Colorectal

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CORRESPONDENCE TO

Indraneil Basu, 25 Llanvanor Road, Golders Green, London NW2 2AR, UK
E: ibasu@doctors.org.uk

Rectal leiomyosarcomas are rare and constitute less than 0.1% of colorectal malignancies.¹ Approximately 290 leiomyosarcomas arising from the rectum have been reported in the literature to date. Although other colorectal malignancies have been associated with irradiation, no firm association exists between rectal leiomyosarcomas and pelvic irradiation. We present the third case of rectal leiomyosarcoma following pelvic irradiation.

Case history

A 79-year-old woman presented to clinic with a history of rectal bleeding, weight loss and altered bowel motions. Clinical examination and rigid sigmoidoscopy were normal. The patient had a past history of endometrial and cervical carcinoma, for which she had undergone an abdominal hysterectomy and received pelvic irradiation 26 years previously. Further investigation with colonoscopy was arranged.

During colonoscopy, a polypoidal rectal tumour with surrounding erythema originating 13cm from the anal margin was identified and biopsied (Fig 1). Histological analysis of the primary biopsy specimens gave no evidence of malignancy. However, due to the suspicious nature of the lesion, the patient was booked for staging computed tomography (CT) and magnetic resonance imaging (MRI) and a repeat sigmoidoscopy, during which further biopsies could be taken.

Imaging revealed the mass to be a 3cm lesion originating 15cm from the anal verge with no clear breach of the muscularis propria and no signs of distant metastases. Histology from the repeat biopsy specimens identified spindle cells demonstrating nuclear pleomorphism. Immunostain-

ing was positive for smooth muscle markers desmin and smooth muscle actin (SMA) but S100 protein, CD34 and CD117 were found to be negative. These findings were in keeping with a benign leiomyoma of the rectum.

Due to the history of previous malignancy, an elective anterior resection was performed rather than the typical

Figure 1 Caecum (A), transverse colon (B), rectal images demonstrating mass (C) and descending colon (D)

wide local excision. The operation was uneventful and the resected specimen sent for further histological analysis. Histological analysis of the full specimen revealed a 3cm by 2.5cm intramural tumour extending 0.5mm beyond the muscularis propria. The tumour demonstrated a fascicular growth pattern with spindle shaped cells demonstrating nuclear atypia and eosinophilic cytoplasm. Brisk mitotic activity was noted throughout the specimen. Again, the tumour cells stained positively for desmin and SMA but negatively for S100, CD34 and CD117. These final findings were consistent with the rare diagnosis of a rectal leiomyosarcoma.

This patient made a slow recovery postoperatively but was discharged home without concern.

Discussion

Mesenchymal tumours of the gastrointestinal tract are uncommon. They are usually found in the stomach and small intestine but can originate throughout the length of the gastrointestinal tract. Those originating from the rectum and anus are unusual. The most common mesenchymal tumours are the gastrointestinal stromal tumours. These possess smooth muscle features on light microscopy but in fact originate from interstitial cells derived from mesenchymal stem cells. To find tumours that actually originate directly from smooth muscle is particularly rare, especially in the anus and rectum.² Such tumours are termed leiomyomas or leiomyosarcomas. Leiomyosarcomas originating from within the rectum constitute less than 0.1% of all colorectal malignancies and approximately 290 cases have been documented in current literature.¹

Both leiomyosarcomas and leiomyomas progress through an asymptomatic intraluminal growth phase before increased size and ulceration lead to a change in bowel habits and bleeding. Less frequently, they can present with tenesmus, pain, obstruction, intussusception and, occasionally, bowel perforation.

Digital rectal examination and sigmoidoscopy often reveal a polypoid or submucosal mass but may only show minor mucosal ulceration. Histological analysis of biopsy specimens helps to diagnose neoplasms and differentiate between benign and malignant growths. CT, MRI and endorectal ultrasound can be used to stage tumours. The preoperative diagnosis is crucial as it determines the treatment strategy. Leiomyosarcomas are commonly treated with an anterior or low anterior resection whereas excisional biopsy is thought to be adequate for their leiomyomas.⁵

Unfortunately, histological analysis of superficial biopsy specimens may not be reflective of the entire tumour mass and leiomyosarcomas can be misdiagnosed as benign leiomyomas. Similarly, malignant potential can also be missed

by CT and MRI. These factors can result in inadequate resections. Of the resected leiomyomas that recur, the majority recur as leiomyosarcomas, suggesting inaccurate initial diagnosis and inadequate resection.⁵ Some studies suggest preoperative histological diagnosis of rectal tumours is adequate in only 29% of cases.⁴ Given that leiomyosarcomas have a poor prognosis compared to their benign counterparts with a five-year survival rate between 20% and 40%, misdiagnosis could significantly affect patient mortality.⁵

Pelvic irradiation is known to be a causative factor in colorectal malignancies but, due to small numbers reported in the literature, no definitive association between pelvic radiotherapy and leiomyosarcomas of the rectum has been made. This is the third documented case of a leiomyosarcoma developing within the rectum following pelvic irradiation. In this case the tumour was discovered 26 years after irradiation, which is consistent with the latent periods of 18 and 22 years documented previously.^{4,5} Although there is insufficient evidence to support an association between radiotherapy and leiomyosarcoma of the rectum, these cases highlight the possibility of association and therefore reinforce the importance of maintaining clinical suspicion in post-radiotherapy patients presenting with similar symptoms.

Conclusions

We would like to stress the importance of long-term follow up of patients who receive pelvic irradiation. Previous advice that surveillance should commence 5–10 years after irradiation seems appropriate for detecting leiomyosarcomas given the long latent periods witnessed in all three reported cases.⁴ We would also like to recommend that clinicians maintain an increased index of suspicion for rectal tumours irrespective of the initial histological and radiological analyses, especially in those with a history of pelvic irradiation.

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