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Rare Rectal Tumour: How to Approach It?

Margarida Rajão Saraiva[a] Daniel Conceição[a] Ricardo Fonseca[b]
Isadora Rosa[a] IPOLFG Colorectal Multidisciplinary Team

[a]Department of Gastroenterology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal; [b]Department of Pathology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal

Keywords
Rectal tumour · Squamous cell carcinoma · Rare diagnosis · Case report

Um caso raro de tumor rectal: como abordar?

Palavras Chave
Tumor do reto · Carcinoma de células escamosas · Diagnóstico raro · Caso-clínico

A 66-year-old Caucasian woman complained of constipation and tenesmus of recent onset. Sole past medical history consisting of a hysterectomy performed 25 years ago due to uterine leiomyomas. Due to her complaints, a colonoscopy was performed and revealed, at 6 cm from the anal margin, a circumferential, friable lesion, with 3 cm of longitudinal extension and an unusual appearance (shown in Fig. 1, 2), with normal mucosa between the distal end of the lesion and the pectinate line. Biopsies were unequivocal for the diagnosis of a squamous cell carcinoma (SCC) (shown in Fig. 3).

The colonoscopy identified no other abnormalities and both the anal canal examination and laboratory evaluations were normal. Magnetic resonance imaging (MRI) confirmed the presence of a circumferential, stenotic tumour of the mid rectum (T4aN2) and a Thoraco-Abdomino-Pelvic Computed Tomography (CT) excluded secondary lesions.

Due to the rare occurrence of a primary rectal SCC (<0.3% of all rectal tumours [1]), especially without any risk factor, an additional multidisciplinary study was carried out to exclude primary occult tumours. This included an upper digestive endoscopy and evaluations by gynaecology, dermatology and oto-rhinolaryngology specialists – all of which ruled out suspicious lesions. A PET-TC-FDG was also performed and only identified the hypermetabolic rectal lesion.

The patient underwent chemoradiotherapy (54 Gy, mitomycin + capecitabine), achieving only a partial response by MRI (mrT3N0), and she was submitted to anterior resection of the rectum. Pathological examination revealed a complete response. Since then, the patient has been under surveillance, with no recurrence after 3 years.

SCC is a condition that, in the gastrointestinal tract, commonly affects the oesophagus or anal canal. In contrast, more than 90% of colorectal tumours are adenocarcinomas. Due to the rare occurrence of rectal SCC, its pathogenesis, risk factors, and optimal

karger@karger.com
www.karger.com/pjg

Correspondence to:
Margarida Rajão Saraiva, margarida.rsaraiva@gmail.com

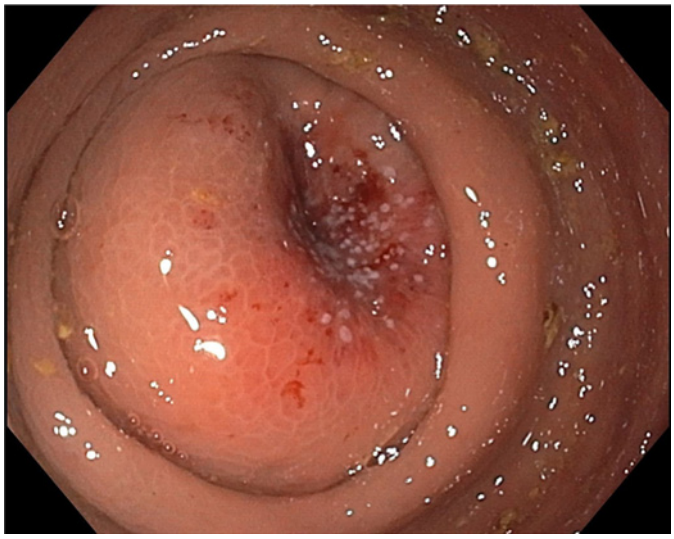

Fig. 1. Endoscopic image of rectal neoplasia.

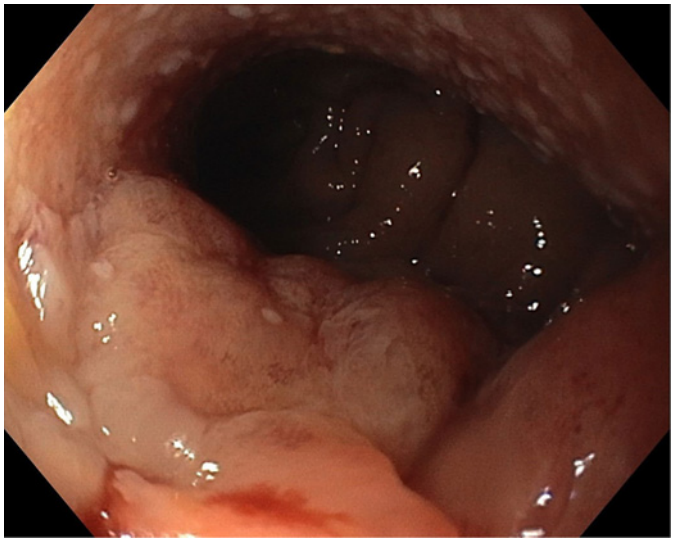

Fig. 2. Endoscopic image of rectal neoplasia.

treatment remain unclear [1] and diagnosis should follow the diagnostic criteria proposed by Williams et al. [2].

Proposed risk factors include: proctitis (ulcerative colitis/parasitic infection), history of radiotherapy, synchronous/metachronous colorectal adenocarcinoma and human papilloma virus infection [3]. While evidence remains limited, the leading theory suggests that chronic inflammation in these patients triggers epithelial changes, leading to squamous metaplasia, dysplasia, and carcinoma.

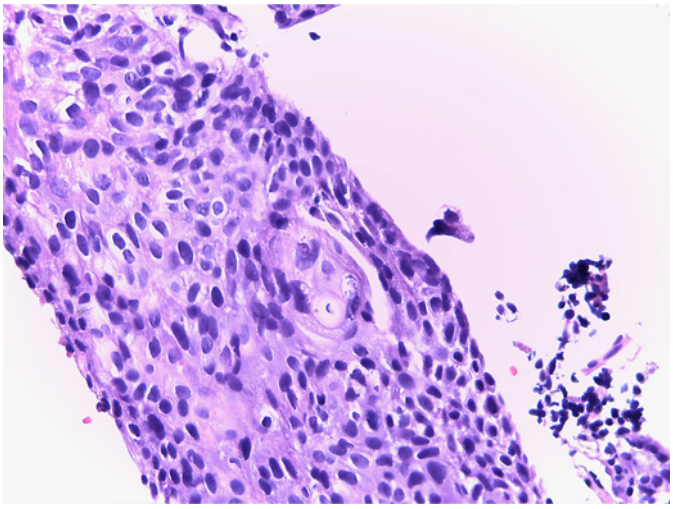

Fig. 3. HE, squamous cell carcinoma. No evidence of glandular differentiation.

In this case, none of the aforementioned risk factors were identified. Although a standardized treatment protocol is missing, chemoradiotherapy has emerged as the preferred treatment for locoregional rectal SCC, in parallel with anal SCC. This has shown better overall survival than surgery (86% vs. 48%), which now plays a salvage role. Rectal SCC has a 5-year survival rate of 48.9%, reflecting a worse prognosis than rectal adenocarcinoma (62.1%) [4].

Due to the lack of specific guidelines, anal SCC surveillance protocols are adopted: clinical and blood tests every 3–6 months for 2 years, then every 6–12 months up to 5 years; pelvic MRI and rectoscopy every 6 months for 3 years; thoraco-abdominal CT/PET-CT annually for 3 years [5]. This case highlights the rare occurrence of rectal SCC and illustrates the importance of a multidisciplinary approach to patient management.

Statement of Ethics

A written informed consent was obtained from the patient for publication of the details of the medical case and accompanying images. This type of manuscript (case report) does not require ethical approval due to local laws.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Margarida Rajão Saraiva wrote the manuscript with support from Daniel Conceição. Ricardo Fonseca performed the pathology examination. All of the members of the Multidisciplinary Colorectal Cancer Group contributed to the management of the patient. Isadora Rosa contributed to the final version of the manuscript.

Data Availability Statement

All data generated or analysed during this study are included in this article and its supplementary material files (for all online suppl. material, see https://doi.org/10.1159/000545592). Further enquiries can be directed to the corresponding author.

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