Primary anorectal melanoma: an unusual cause of rectal bleeding

A 83-year-old man was referred to our institution complaining of painless rectal bleeding and a protruding mass during defecation. Digital rectal examination confirmed a friable soft tissue mass. Clinical history and remainder physical examination were unremarkable. Blood investigation revealed microcytic hypochromic anaemia. Carcinoembryonic antigen level was within the normal range.

Imaging Findings:
Endoscopic ultrasonography (EUS) revealed a solid, homogeneous and hypoechoic tumour in the anorectal junction (Fig. 1). Due to the patient's painful, it was not possible to cross the tumour with the probe. The tumour invaded the submucosa, but no muscular invasion or mesorectal lymph node metastases were observed.
Magnetic Resonance Imaging (MRI) performed for further characterization and local staging, confirmed a solid polypoid lesion in the anorectal junction. The hypointense line corresponding to muscularis propria was intact and no internal sphincter invasion was observed. The tumour demonstrated high signal intensity on T1-weighted images and low signal intensity on T2-weighted images. On diffusion-weighted imaging (DWI) the tumour demonstrated restricted diffusion, represented by high signal intensity on b 1000 s/mm² and corresponding low signal on apparent diffusion coefficient (ADC) map.

Discussion:
Primary anorectal melanoma is a rare malignancy comprising less than 1% of all melanomas and 0.25-1.25% of all anorectal malignancies. The disease typically affects Caucasians between the fifth and eight decades of life and its incidence has doubled in the past 20 years in Western countries. The prognosis remains poor with a five-year survival rate of 10-15% [1, 2].
Rectal bleeding is the most common presentation but non-specific symptoms such as anal pain, tenesmus or pruritus are frequently referred. The disease is confused with benign entities (haemorrhoids or polyps) and the diagnosis is usually delayed [1-3].
The pathophysiology of primary anorectal melanoma remains unclear. It arises from melanocytes present in the mucosa and some studies suggest that there is a role of activating KIT proto-oncogene receptor tyrosine kinase in its pathogenesis [3].
Primary anorectal melanoma is more commonly located in the anal canal or close to pectinate line, extending along
the submucosal plane proximally towards rectum, simulating a primary rectal tumour. Rarely, tumours arise in the rectum itself. The evaluation of the primitivity includes the presence of a single lesion, the absence of other primary locations and the absence of previous removal of melanoma or atypical melanocytic lesions [4]. Different imaging techniques are used to evaluate primary anorectal melanoma, metastasis and treatment responses. EUS is helpful to differentiate between solid and cystic lesions and to evaluate the lesion size, location and depth of bowel wall invasion. MRI is widely used as the diagnostic modality for anorectal abnormalities and is recommend for local staging of malignancies. On MRI, anorectal melanoma usually presents as an intraluminal polypoid mass that not causes obstruction. Due to its paramagnetic feature, the melanotic component of melanomas shorts the T1 relaxation time and increase the T2 relaxation time, which results in high signal intensity on T1-weighted images and low signal intensity on T2-weighted images. These tumours usually show moderate to strong enhancement and diffusion restriction. Positron emission tomography/computed tomography is recommended for systemic staging and for response assessment due to its high sensitivity and specificity [2, 5]. Surgery remains the mainstay of treatment and includes wide local excision, endoscopic mucosal resection or abdomino-perineal resection. Sentinel lymph node procedure is recommended before inguinal lymph node dissection, although it is not predictive of prognosis. Chemotherapy, radiation therapy and immune therapy have limited role [6].

The Editorial Board has modified data in this case to guarantee patient anonymity, and ensured that those changes do not alter the scientific value.

**Differential Diagnosis List:** Primary anorectal melanoma, Haemorrhoids, Benign polyp, Anal squamous cell carcinoma, Rectal adenocarcinoma

**Final Diagnosis:** Primary anorectal melanoma

**References:**


Description: Endoscopic ultrasonography shows a homogenous and hypoechoic solid tumour (asterisk) based in the right posterolateral distal rectal wall. Muscularis propria is intact (arrows). **Origin:** Department of Radiology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal.
Figure 2

Description: Axial T1-weighted image shows a high signal intense polypoid tumour (asterisk) in distal rectum, just above anal verge. Origin: Department of Radiology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal
Description: Axial-oblique T2-weighted image shows a clearly defined mass with low signal intensity (asterisk). The hypointense line corresponding to muscularis propria is intact (arrows) and no perirectal infiltration is seen. Origin: Department of Radiology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal
Description: Diffusion-weighted image confirms a high cellular lesion with high signal intensity on b 1000 s/mm² (arrow). Origin: Department of Radiology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal
Description: The lesion has low signal on the corresponding ADC map (asterisk). Origin: Department of Radiology, Instituto Português de Oncologia de Lisboa Francisco Gentil, Lisboa, Portugal