



An Unusual Polyp: Rectal Melanoma

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Abstract

Primary rectal melanoma is a rare disease that requires a high index of clinical suspicion when faced with unusual colonoscopic findings. We present the case of a 67-year-old gentleman who had a primary rectal melanoma discovered on a colonoscopy to investigate bright rectal bleeding. No other sources of primary disease were discovered clinically or radiologically pre-operatively suggesting that this was a true site of primary disease. He went on to have an abdomino-perineal resection and made an uneventful recovery.

Introduction

Primary rectal melanoma is rare anal squamous cell carcinoma and rectal adenocarcinoma being far more prevalent [1]. Likewise melanomas are far more likely to arise from anal mucosa than rectal mucosa although exact proportions are difficult to ascertain given the rarity of this disease entity. Of all primary melanomas less than 1% is ano-rectal in origin [2]. The gastrointestinal tract is far more likely to be the site of secondary disease from primary cutaneous melanomas. Surgery remains the gold-standard treatment although newer adjuvant treatments relating to B-RAF mutations currently being used in the treatment of cutaneous melanomas may have an increasing role in the future [3]. Despite aggressive surgical intervention the prognosis remains poor with a 5 year survival of less than 20% [4].

Case Presentation

A 67-year-old caucasian male presented with a 6 week history of bright red rectal bleeding and occasional anorectal discomfort associated with defecation. He had trialled an over the counter topical haemorrhoid cream without improvement. His past medical history included obesity, hypertension, stage III chronic kidney disease, type 2 diabetes mellitus and psoriatic arthritis for which he was receiving methotrexate. There was no personal history of cutaneous or non-cutaneous malignancies, and there was no family history of premature malignancy. Abdominal examination was unremarkable. A rectal examination performed at the time of colonoscopy was positive for a palpable rectal mass. The distal extent of the mass was 4-5cm from the anal verge. Colonoscopy revealed a 35mm polypoid mass with surrounding pigmented mucosa (Figure 1). This was removed with endoscopic mucosal resection (EMR)/hot snare. Histology revealed a spindle cell tumour with variable amounts of brown pigment (Figure 2A and B). Immunostaining was positive for S-100 (Figure 2C) and CD117 (Figure 2D), whilst negative for AE1/AE3, desmin and BRAF. Up to 48% of melanoma cases can show positivity to CD117 [5]. A strongly positive stain for S-100 in addition to melanin pigmentation confirmed the diagnosis of malignant melanoma. A follow up colonoscopy revealed residual mucosal pigmentation surrounding the previous resection site, with biopsies confirming the presence of residual melanoma. Further molecular pathology testing for BRAF and NRAS was performed and found to be negative. A computed tomography scan of the chest, abdomen and pelvis, rectal MRI scan and PET scan showed no radiological evidence of locoregional nodal or distal metastatic disease, and no other likely sites for primary disease. The tumour was staged on MRI as T2N0. Dermatological review pre-operatively failed to identify any suspicious pigmented skin lesions to suggest that the rectal melanoma could have been a secondary deposit. Surgical intervention followed with the patient undergoing laparoscopic extralevator abdomino-perineal resection (Figure 3). Histology confirmed an R0 resection with grade 3 mesorectal dissection. The tumour had not extended beyond the muscularis propria (T2) and 0 of the 16 nodes were involved

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Received Date: 09 Mar 2016

Accepted Date: 20 Apr 2017

Published Date: 11 Oct 2017

Citation:

Chakradeo K, Cross T, Lampe G, Nabi H, Safa S. An Unusual Polyp: Rectal Melanoma. *Clin Surg*. 2017; 2: 1670.

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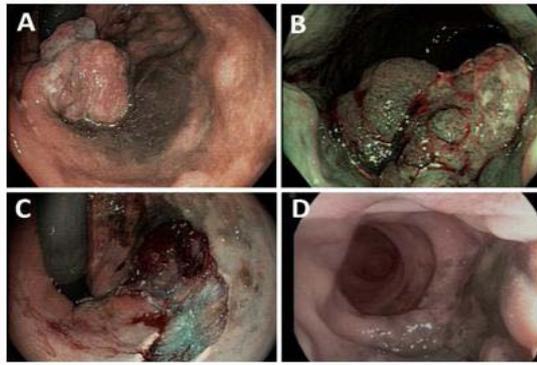


Figure 1: (A) Rectal mass with surrounding mucosal pigmentation is visualised on colonoscopy with white light and (B) narrow band imaging. (C) Post polypectomy site on retroflexion. (D) Follow up colonoscopy taken 2 weeks after initial procedure with evidence of mucosal pigmentation at previous polypectomy site.

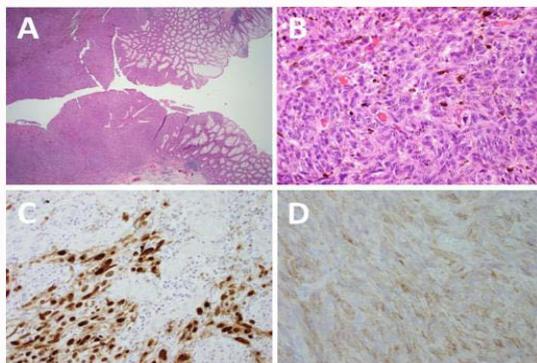


Figure 2: (A) Histology low power shows colorectal mucosa infiltrated by lesional tissue, with a hint of melanin pigment at the upper section. (B) Histology medium power shows pleomorphic spindle cells, prominent nucleoli and brown (melanin) pigment. (C) Immunostain positive for S-100. (D) Immunostain positive for CD117.

(N0). After an uneventful recovery he was discharged at day 3 post-op with an outpatient oncology follow up referral (given his BRAF negative status, no adjuvant therapy was offered). On follow-up at 6 months post-op he remains clinically and radiologically disease free.

Discussion

Anorectal malignant melanoma is a rare and unusual cause of rectal bleeding. Only 0.05% of all malignant colorectal diseases are found to be melanoma [1] and around 1% of all melanomas are located in the anorectal region [2,3]. Rectal melanoma has a poor prognosis with 5-year survival of less than 20% [1,3-5]. Rectal melanomas can be pigmented, as in this case, although can also present as a non-pigmented lesion. Immunohistochemistry should be performed in all suspicious cases and is especially helpful in distinguishing non-pigmented melanoma [6,7]. Surgical intervention remains the treatment of choice and is associated with improved survival. Although our patients' tumour was negative for BRAF, identification of BRAF in tumours is important to allow for possible targeted chemotherapy regimens. Additionally, it is important to recognise that immunosuppression with agents such as methotrexate may play a role in increasing the risk of malignancies including melanoma. This

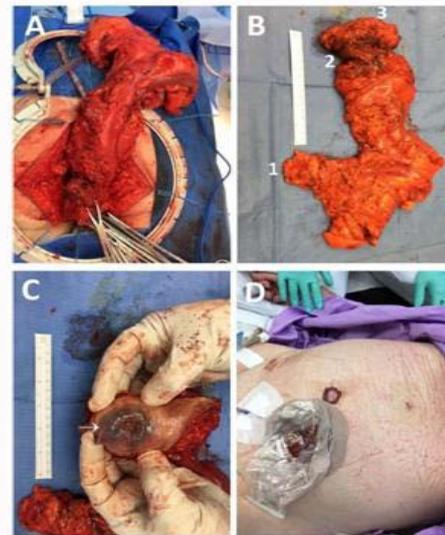


Figure 3: (A) Intra operative photo of the prone extralevator perineal dissection with the resected specimen about to be extracted from the perineum. (B) Resected specimen showing high tie on the inferior mesenteric artery pedicle (1), levator ani muscles in the extra levator resection (2) and the perianal skin (3). (C) The perianal skin of the resected specimen with the pigmented skin extending from the lesion (ARROW). (D) Immediately post op. the patient has an end colostomy and a completed laparoscopic procedure.

case highlights the importance of further investigation in patients with mild rectal bleeding that can be mistaken for haemorrhoids or other less serious aetiologies. Although rectal melanoma is an uncommon cause of rectal bleeding, early diagnosis and management is crucial to optimise patient outcome.

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