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Melanoma of the Anorectum: A Rare Entity - Case Report with Review of Literature

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Abstract

An uncommon form of malignant melanoma is anorectal malignant Melanoma (ARMM), attributing to only 1% of all rectal malignancies. About There are times when the diagnosis appears straight forward, and at other times it is confusing thereby requiring a high degree of suspicion on the part of the treating physician. Due to the aggressive nature of this disease, an early diagnosis and prompt treatment are essential. Specific mutations in the c-Kit/stem cell factor (SCF) pathway, the endothelin receptor type B/endothelin pathway, and the Sox10transcription factor are being incriminated in the pathogenesis pigment and enteric nervous system disorders including melanomas. This may form a basis for further research and treatment of these disorders.

Keywords: Anorectal Melanoma; aggressive; Ckit/stem cell factor (SCF) pathway

Introduction

Only 1% of all anorectal carcinomas are melanomas typically presenting in the fifth or sixth decade of life and predominantly in women. Due to the aggressive nature of this disease, an early diagnosis and prompt treatment are essential. The anorectum is the third most common location of malignant melanoma after the skin and retina [1, 2]. Unlike other forms, there is no association with exposure to ultraviolet light. Diagnosis is often delayed and a poor prognosis is compounded by the aggressive nature of the malignancy resulting in a median survival of 24 months and 5-year survival in only 15% of cases [3] As a consequence, few surgical guidelines are available.

Case History

The patient was a 65-year-old female who presented to the out-patient department with a six-month history of bleeding per rectum. Abdominal examination revealed no abnormality. There was no significant palpable inguinal lymphadenopathy. Per-rectal examination revealed a blackish discolored polyp of the peri-anal skin circumferentially around the anal orifice, extending for about a centimeter away from the anal orifice. Her hematological and biochemical parameters were all within normal limits.

This polyp was resected and histopathological examination showed the characteristics of malignant melanoma. The Immunohistochemistry results showed positive expression of S100. Fig 1(a) Histopathology of the polyp showed an ulcerated mucosal lining with underlying spindloid cells in long fascicles with pleomorphism and focally prominent nucleoli. Brownish melanin pigment was see both intra and extracellulary. Fig 1(b) These cells showed membranous positivity for S100.

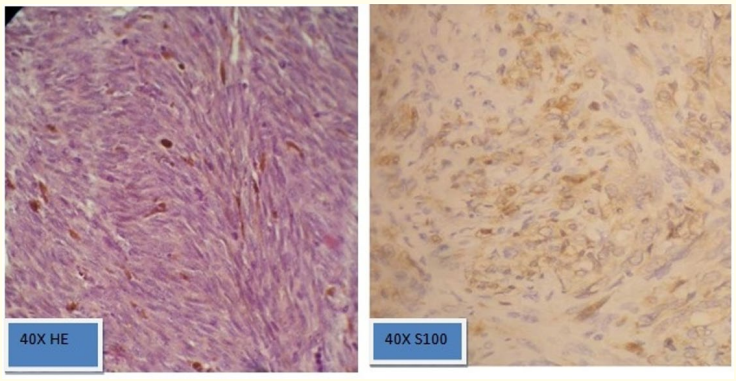

Figure 1: HE stained spindley cells in long fascicles with pleomorphism and focally prominent nucleoli. Brownish melanin pigment was see both intra and extracellulary.
Figure 1(b): These cells showed membranous positivity for S100.

Discussion

The majority of colorectal and anal malignancies are adenocarcinomas and squamous cell cancers. Despite the predominance of these neoplasms at these locations, rare histotypes of the colon, rectum, and anus occur. These histotypes include lymphoma, melanoma, diffuse cavernous hemangioma, and sarcomas, such as leiomyosarcoma or Kaposi's sarcoma. Malignant melanoma of the anal canal is an extremely rare and lethal tumor. The first case of anal melanoma was reported by Moore in 1857 [1, 2]. Malignant melanoma is the fifth leading cause of new cancer diagnosis in males in the and the sixth in females worldwide [3]. This incidence is slowly rising by approximately 3% per year. These increases may be attributed to increased screening and codification of pathologic standards for diagnosis.

Patients present themselves with local symptoms like rectal bleeding and a changed detection pattern [1-6]. At the Memorial Sloan Kettering Cancer Centre, 85 patients were seen in a 64 year period between 1929 to 1993. Survival was poor with a 17% five year survival with a median of 19 months [4]. Besides occurring in the anal canal, melanomas most frequently originate from the mucous membranes lining the respiratory, digestive, and genitourinary tracts or in the eyes as well as in the cerebral meninges [4]. Females are more likely to be affected, generally presenting in the sixth or seventh decade [5, 6]. Clinically, the patients present with bleeding per rectum or diarrhoea with tenesmus. The growth may ulcerate giving rise to severe pain on defecation. On examination, a tumour can be seen and can always be palpated, although on occasion the primary growth is small. Patchy perianal pigmentation may be noticed. Differential diagnoses include hemorrhoids, carcinoma of the rectum and prolapsing rectal polyp which was one of the differentials in our case along with prolapsed hemorrhoids. This requires a high degree of suspicion to diagnose these tumours early.

The lesions can be located in the anal canal, rectum or both with the majority of them arising from the dentate line of the anal canal. They tend to spread submucosally, and by the time they cause symptoms the extent of invasion is usually beyond surgical cure [7, 8].

There are times when the diagnosis appears straight forward, and at other times it is confusing thereby requiring a high degree of suspicion on the part of the treating physician. The abundant lymphatics of the anorectum probably facilitate the high rate of inguinal and iliac lymph node metastastes [9-11]. The rich vascular network in this area promotes hematogenous spread to liver, lung, bone, brain, and other organs. In addition, anorectal melanomas often achieve large size and nodular growth before clinical detection [10].

The Clark level and Breslow index used for evaluation of cutaneous melanomas are not applicable to extracutanous melanomas and, at present, there are no consistent, internationally accepted therapy standards for this form of the disease [11].

If a biopsy shows a specimen suspicious for sarcoma (e.g. leiomyosarcoma), one should be alert. Preferably, S-100 staining should be performed in addition to routine staining. A positive S-100 stain suggests the tumour most likely to be a melanoma. Subsequently dissemination studies, including chest X-ray and CT-scan of chest/abdomen/pelvis are performed. Curative surgical resection has to be performed in the absence of metastases proven by dissemination studies.

Considerable dispute exists regarding the cell of origin for Anorectal malignant melanoma. While the presence of melanocytes has been relatively well described in the mucosa of the head and neck [13, 14] and the esophagus [12] its presence in the intestinal mucosa from the stomach to distal rectum is not well explained and not much has been written about it [11, 12].

Melanocytes are usually found below the dentate line and thus it has been hypothesized by various studies that anorectal melanomas are actually derived from these normal melanocytes distal to the dentate line [9]. Certain studies with Immunohistochemical stains have shown the presence of melanocytes above the dentate line, HMB-45 and S100 [8-10]. This has led to the conclusion that ARMM can arise directly from melanocytes located in the intestinal epithelium of the proximal anus or distant rectum hence this is referred to as primary anorectal melanoma. Embryologically, cells migrate from the neural crest to enter the dorsolateral pathway differentiate into melanocytes and eventually populate their sites of colonization, while cells entering the ventral pathway are the neurogenic precursors of the peripheral and enteric nervous system.

Neural crest cells are typified by a characteristic set of transcription factors including *Snail2* (*Slug*), *Sox10*, *FoxD3*, and *Sox9* [5, 9, 10]. Melanocyte migration and differentiation involves a complex interplay of cell signaling pathways Specific mutations in the *c-Kit/stem cell factor* (*SCF*) pathway, the *endothelin receptor type B/endothelin* pathway, and the *Sox10* transcription factor pathway being associated with a variety of related pigment and enteric nervous system disorders including Waardenburg syndrome, and Hirschprung's disease [11].

In conclusion, anorectal melanoma is a rare and aggressive disease. Because of nonspecific symptoms they can be easily mistaken for hemorrhoids. Furthermore, the prognosis depends on the staging, and it is important to detect anorectal melanoma at an early stage [13].

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