Leiomyosarcoma of rectum and anal canal - a diagnostic challenge: A review

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Leiomyosarcoma of rectum and anal canal-a diagnostic challenge: A review

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Abstract

Leiomyosarcoma of the rectum and anal canal is a rare disease. Diagnosis is virtually impossible without proper immunohistochemistry. A 50 year old woman was incidentally found to have a rectal nodule which initially turned out to be a malignant melanoma. The patient later returned with a large rectal growth for which she underwent an abdominoperineal resection. She was diagnosed as having leiomyosarcoma based on the immunohistochemistry results

KEYWORDS: rectal leiomyosarcoma, anal leiomyosarcoma, calponin positivity, rectal leiomyosarcoma treatment
Introduction:

Leiomyosarcoma of the rectum and anal canal is a rare disease. There are only 18 reported cases of anal canal leiomyosarcoma and around 300 reported cases of rectal leiomyosarcoma. Diagnosis is virtually impossible without proper immunohistochemistry. The natural history and appropriate treatment protocols have not been well defined due to a scarcity of such cases and their details.

Case report

A 50 year old female patient had undergone a vaginal hysterectomy for uterine prolapse in June, 2004. During the operation an incidental hard nodule (1x1cm) was found on the anal canal which was excised immediately. Histopathology revealed a malignant melanoma with free margins. Clinical examination, digital rectal examination, a colonoscopy and a CT scan of the abdomen revealed no abnormalities. Even after six months a sigmoidoscopy (November, 2004) suggested that the rectum was normal. However a year later in September, 2005 she developed a nodular lesion (2x2cm) near her anal verge. A biopsy, at first, revealed a fibroepithelial polyp but an excision biopsy done without injuring the sphincter, suggested that it was a neurofibroma. She was regularly followed till November 2006. After that she was lost to follow up. One year later she then presented to our Department with difficulty in passing stools and anaemia. Clinically she was found to have a large growth involving the rectum, anal canal, anal sphincter...
complex and invading the posterior wall of the vagina which was confirmed by a CT scan of the abdomen and pelvis (Fig 1). An incisional biopsy suggested a malignant nerve cell sheath tumour. We embarked upon an abdominoperineal resection as the sphincter complex was involved. The complete mass was removed along with the posterior wall of the vagina which was reconstructed. The growth was a large polypoid tumour 8.5 cms in its greatest axis involving the rectum and anal canal. Ten lymph nodes were found in the pararectal fatty tissue none of which harboured any metastasis. The surgical margins of excision were free. On microscopy sections showed a tumour composed of interlacing fascicles of spindle shaped cells with elongated, plump, wavy nuclei. At places the spindle cells were arranged in a whorled pattern and the cellular areas showed 2-3 mitotic figures per 10 high power fields (Fig 2).

This picture can be seen in spindle cell sarcoma, malignant peripheral nerve sheath tumour or leiomyosarcoma. Immunohistochemistry was done to differentiate them. The tumour was positive for smooth muscle actin and calponin but was negative for c-kit, CD34, HMB-45, S-100, desmin and cytokeratin. Based on these a diagnosis of low grade leimyosarcoma was made. Post operatively she received six cycles of chemotherapy which included cyclophosphamide, doxorubicin, vincristine and dacarbazine. She is on regular follow up and is doing well with no evidence of a recurrence.

Discussion:
Leiomyosarcomas are malignant mesenchymal tumours arising from smooth muscle cells and account for 10-20% of all soft tissue sarcomas. They are more common in females. They usual sites affected are the skin, extremities and retroperitoneum. They are the commonest of all gastrointestinal sarcomas, but account for less than 0.1 to 0.3 per cent of all malignancies. True rectal leiomyosarcomas are rare and account for less than 0.1% of all malignant tumors of the rectum. They tend to occur between 50 to 69 years of age, and approximately 20% of rectal leiomyosarcomas reported from 1881 to 1996 had metastasized at the time of diagnosis.

Leiomyosarcoma presents with a diagnostic dilemma especially on haematoxylin and eosin stained sections as it mimics a number of conditions like leiomyoma, gastrointestinal stromal tumours, tumours of the nerve sheath, malignant melanoma of spindle cell as well as spindle cell carcinoma. Preoperative diagnosis is difficult to achieve and evidence suggests that adequate pre-operative histological diagnosis was found in only 29% cases. As a biopsy does not include the whole mass actual characterization especially malignant evaluation is often only possible after a complete excision of the mass. Leiomyosarcoma is characterized by malignant spindle cells with cigarshaped nuclei arranged in interweaving fascicles. They often have an increased amount of nuclear and cellular atypia, some degree of necrosis, and a high number of mitoses.
Immunohistochemistry plays an important role in identifying and differentiating these similar appearing pathologies. Leiomyosarcoma classically stain with, smooth muscle actin, vimentin and desmin\(^3\) with smooth muscle actin positivity being present in 100% cases.\(^7\) Basic calponin is a recently discovered marker which is an actin, tropomyosin and calmodulin-binding protein originally\(^8\) isolated from smooth muscle and is found in 75% of cases with Leiomyosarcoma\(^7\). Vimentin is likely to be positive in all these tumours and it would not help in differentiating them. Desmin is more of a skeletal muscle marker and so it can be negative in leiomyosarcoma which has a smooth muscle origin. Based on smooth actin and calponin positivity this patient was diagnosed as leiomyosarcoma. S-100 negativity excludes malignant peripheral nerve sheath tumor which is the malignant counterpart to benign soft tissue tumors such as neurofibromas and schwannomas. HMB-45 is 100% specific and 93% sensitive for malignant melanoma\(^9\) and a negative HMB-45 and S-100 effectively rules out melanoma. Gastrointestinal stromal tumours(GIST) are mesenchymal tumours arising from the interstitial cells of Cajal characterized by c-kit positivity which closely resemble leiomyosarcomas. In fact historically many GISTs have been wrongly diagnosed as the latter. Here, GIST was excluded due to negative c-kit and CD 34 expression. Further evidence for diagnosis may be found on electron microscopy but this is not widely available for diagnostic purposes. Furthermore, although the diagnostic challenges lie with the pathologist, these are very similar tumours in behaviour characterized by rare lymph node involvement\(^10\) but high recurrence rates. So irrespective of histology, treatment in these tumours is always surgical and therefore not a dilemma to surgeons.\(^11\)
Here the patient first had a lesion which histology identified as malignant melanoma and was accordingly treated. The second time a diagnosis of neurofibroma was made. The histology pictures on paraffin blocks of these tumours can be frustratingly similar. Whether these tumours are precursors to leiomyosarcoma or a diagnostic mistake is best left to speculation. The rarity of leiomyosarcoma in these sites precludes discovery of any obvious evolution of these tumours.

There lies a lot of debate over treatment of melanomas of the anal canal with some, like in Memorial Sloan Kettering suggesting abdominoperineal resection (APR) for even small lesions while others believe a local excision would suffice and that APR does not improve survival. Therefore as the initial lesion was around 1 cm in diameter and the excision had clear margins we did not perform a radical procedure.

While there is no actual consensus about the type of surgery for rectal or anal canal tumours a radical procedure like low anterior resection or APR is usually preferred. However, in this case, anal sphincter involvement makes APR the only viable option. Chemotherapy and radiotherapy have no role but adjuvant therapy may produce some results. Recurrence rates are very high. It is more than 80% exceeding the propensity of leiomyosarcomas in other areas of the gastrointestinal tract to recur. Hence, a prolonged follow up is required. A five year survival rate is around 20% to 25%. Prognosis is poor and depends on tumour size, histological grade, mitotic index and local staging. But a dearth of such cases limits actual understanding of progression and behaviour of these tumours.
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