Case report

Can a perineal mass be a leiomyosarcoma? An interesting case

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Abstract: A sarcoma is not so easily recognized due to its wide spectrum of presentation. We describe a rare case of leiomyosarcoma of the perineum which presented as a recurring mass in a young woman, and the plan of management that has been provided. The leiomyosarcoma is one of the rarest varieties of sarcoma. Generally it is observed in the fifth or sixth decade of life. The common site of occurrence is the uterus, and the plan of management is very different compared to other sites. The present case is that of a recurrent perineal mass which was not suspected to be malignant prior to the initial surgery. The patient received chemotherapy once the lesion was diagnosed as rhabdomyosarcoma based on pathology report, immunohistochemistry reports were inconclusive. Recurrance while on chemotherapy lead to change in treatment plan Abdomino-perineal resection was not acceptable to the patient. A treatment plan was decided upon to tailor to the patients needs, a sandwich therapy was decided and pre-operative radiotherapy followed by surgery and adjuvant chemotherapy were given. Patient did not have symptoms of recurrence with a regular three month follow up for one year.

Key words: Adjuvant chemotherapy; Immunohistochemistry; Perineal leiomyosarcoma; Recurrent perineal mass.

INTRODUCTION

Leiomyosarcoma is a relatively rare tumor, which presents in the fifth or sixth decade of life.[1,2] This malignant smooth muscle tumor arises from the wall of the gastrointestinal tract, uterus, soft tissues, and retroperitoneal tissue. It comprises less than 0.1% of all ano-rectal malignancies. The definitive treatment plan depends on the site of tumor, age of patient and recurrence rate. Sixty percent recurrence rate after primary surgery is noted in the literature.[3]

CASE REPORT

A 23 yr old woman presented with a perineal mass (5.7x4.1x5.7cm) which recurred one month after the primary excision at the same site. The initial excision surgery was performed in another hospital where it was diagnosed as a rhabdomyosarcoma on histopathology basis and received chemotherapy with vincristin, adriamycin and cyclophosphamide. In view of progression of tumor despite of two cycles of chemotherapy the case was refered to our center. The revised histology was inconclusive therefore immunohistochemistry was performed which was positive for smooth muscle actin with cytoplasmic activity, negative for desmin, myogenin and s-100. A CT scan of the abdomen and pelvis demonstrated a lesion of 11.4x12.5x15.3cm. On biopsies taken in the recurred lesion immunohistochemistry was positive for desmin, strongly postitve for calponin and had no reaction for CD99 and HMB 45 (Figure 1), therefore it was unlikely to be a rhabdomyosarcoma. The histological features were suggestive of an undifferentiated sarcoma with round to ovoid cells and cigar shaped nuclei, and a final diagnosis was given as "perineal leiomyosarcoma".

In view of the large size of the lesion, an abdomino-perineal resection was planned, but the young patient did not give her consent and after a literature review, a more suitable option was offered.

A 50Gy irradiation in 25 fractions to the whole pelvis and perineum produced a regression of the tumor to 9x9x8cm. On clinical examination a firm swelling in the right side of the perineum with the previous scar were noted. At rectal examination a bulge in the antero-lateral walls of the anal canal could be felt but there was no growth within the anal canal. Informed consent was obtained for a wide excision of the tumor, abdomino-perineal resection being a second option. At surgery (Figure. 2), under general anesthesia, the proximity to postero-lateral vaginal wall and anterior rectal wall were noted. Incision was given keeping 1cm margin from the visible mass, the tumour was excised en bloc. After a wide excision, approximation of skin was achieved successfully. The post-operative period was uneventful, the patient was discharged on the fourth post-operative day. Histopathology reported a diagnosis of leiomyosarcoma with tumor free margins. Six more cycles of ifosfamide, adriamycin and mesna were given successfully . Till last follow up visit patient did not have any complaints of incontinence of flatus or stool, nor any visible or symptomatic recurrence after 12 months of surgery.

DISCUSSION

In the literature perineal leiomyosarcoma is an extremely rare tumour. Among all soft tissue sarcomas 5-10% are leiomyosarcomas.[4] Soft tissue leiomyosarcoma is thought to arise from the smooth muscle cells lining the small blood vessels, usually from the gastrointestinal tract and the uterus, but perineum as primary site is extremely rare and more aggressive than other superficial leiomyosarcoma.[4] Most of the time it presents as a painless, nontender mass and a benign histopathology is expected. It affects women more than men (2:1),[1,2] usually in the 5th and 6th decades of life.[4] The gender distribution has also been attributed to proliferation of smooth muscle in response to estrogens.[2] In a woman with complaint of a slow growing mass on the perineum the initial clinical differential diagnoses are Bartholin abcess, perineal abcesses or leiomyoma.

Histologically leiomyosarcoma has been divided into two subgroups: the superficial dermal form of leiomyosarcoma is thought to arise from the arrector pili muscles, and the deep subcutaneous smooth muscles of vascular wall.

Though histologically they may be same but treatment plan and prognosis differs in various leiomyosarcomas. For treatment purposes they are divided into four groups:[5,6]

Leiomyosarcoma of retroperitoneal somatic soft tissue.

Leiomyosarcoma of cutaneous origin.

Leiomyosarcoma of vascular origin

Leiomyosarcoma in immunocompromised host.

The tumor is composed of highly cellular fascicles of spindle-shaped cells. The cells have nuclei that are elongated and blunt ended giving typical "cigar" appearance. The degree of differentiation may vary within a single tumor itself. Classical immunophenotyping of leiomyosarcoma includes positive vimentin, desmin and smooth muscle actin (SMA) staining.[5] Desmin staining is sensitive and specific but cannot differentiate between rhabdomyosarcoma and leiomyosarcoma. Another histological differential diagno-

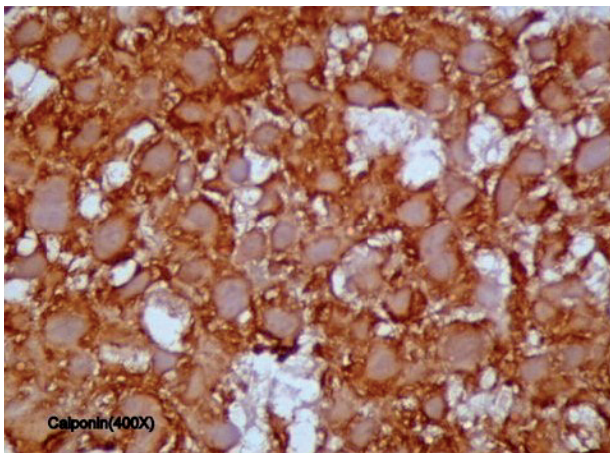

Figure 1. - Immunohistochemistry imaging, colponin postive.

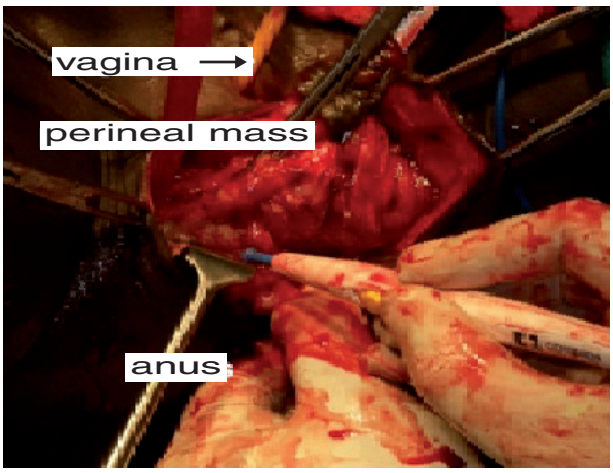

Figure 2. - Intraoperative picture of wide excision of perineal leiomyosarcoma: arrow showing intact vagina, dissected mass and intact anus

sis can be with spindle cell rhabdomyosarcoma which gives positive immunohistochemistry results for SMA and myogenin but no reaction to h-caldesmon which is seen reactive only in smooth muscle cell. Since the present case was SMA positive and myogenin negative it did not require h-caldesmon to differentiate its smooth muscle origin and spindle cell differentiation.To designate a tumor as rhabdomyosarcoma, myogenin should be positive.[5] Therefore rhabdomyosarcoma can be ruled out in this case.

This tumor has a variable prognosis that depends on its location, and treatment differs accordingly. Surgery is the definitive treatment of choice for perineal leiomyosarcoma to avoid metastases. The best approach would be abdomino-perineal resection due to the aggressive nature and the high recurrence rate.[7] Grobmyer et al reported nine cases who received adjuvant external beam radiation following wide excision. The lesions were high grade, up to 5 cm, and recurrence was seen in 60% of the cases. They also reported a median survival of 54 months which seems promising in this disease.[3]

The 50Gy radiation treatment in our patient shrinked the tumor and made it locally resectable avoiding the abdomino-perineal resection.

The chemotherapy agents used in sarcoma include doxorubicin and Ifosfamide, gemcitabine and taxotere (docetexel), dacarbazine and ecteinascidin.[2] Currently ifosfamide is found to be active in sarcomas that have failed to respond to doxorubicin-based regimen, as well as in recurrent or metastatic cases. The results of randomized control trials show that response rate to doxorubicin with or without ifosfamide are significantly higher than the ifosfamide-containing arm.[8] We have opted for ifosfamide and doxorubicin, that have shown promising results.

The median time from treatment to first recurrence noted was 21 months in a previous study.[3] The mean interval of metastases is 31 month (0-12 yr) detected over 5 years after diagnosis of the primary tumor and 8 years in a case report.[9] The earliest death after initial surgery due to recurrence/metastases has been reported by 16 months post surgery. The present case has not been been found with any recurrence till her last follow up 12 months after surgery. Leiomyosarcoma cases need a follow up every three months for the first two years and every six months for the next three years.

CONCLUSION

The perineal leiomyosarcoma in a young woman is rare. Immunohistochemistry is diagnostic when histopatholgy is doubtful. Rhabdomyosarcoma is common in young age and leiomyosarcoma should be considered in the differential diagnosis of asymptomatic perineal mass.Neoadjuvant radiotherapy in a large tumor may make resectable an unresectable disease.

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