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Case Report

Low grade fibromyxoid sarcoma of the perineum: a surgical challenge

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ABSTRACT

Low-grade fibromyxoid sarcoma is a rare, slow growing and deceptively benign-appearing neoplasm. It is presently diagnosed on the basis of histopathology (typical fibro-myxoid appearance), immunohistochemistry (vimentin staining) and cytogenetics [chimeric FUS/CREB3L2 gene produced by t (7;16), (q33;p11)]. This tumour should be differentiated from other resembling tumours like myxofibrosarcoma, sclerosing epitheloid fibrosarcoma, desmoid fibromatosis and others. It has the potential for local recurrence and late metastasis, if not treated adequately. Although surgical excision is the only hope for treatment, reporting of details of surgical management have often been neglected for this primarily pathological entity. We report a case of a 30-year female who had a perineal mass which was managed by wide surgical excision. It was diagnosed as low-grade fibromyxoid sarcoma on the basis of histopathology and immunohistochemistry. This extremely rare presentation is, thus, discussed in context of challenging nature of its surgical excision. Patient is in follow-up and has no recurrence even after 5 years.

Keywords: Deceptively benign, Immunohistochemistry, Late metastasis, Local recurrence, Low grade fibromyxoid sarcoma, LGFMS, Perineum, Wide surgical excision

INTRODUCTION

Low-grade fibromyxoid sarcoma (LGFMS) is a rare soft tissue tumour of fibroblastic origin.[1] It is a slow growing, deceptively benign-appearing but potentially malignant neoplasm.[2] It shows an aggressive course in the form of, not uncommon, local recurrences and late metastases, if not treated adequately.[2] It is now considered a separate pathological entity, first reported by Harry Evans in 1987, so also sometimes referred to as evans tumor.[1] The characteristic histopathology, immunohistochemistry (IHC) and cytogenetics help in differentiating it from other rare soft tissue sarcomas. PubMed search yielded the fact that perineal presentation of this tumour is exceedingly rare, as it is commonly found in superficial or deep locations in the extremities.

CASE REPORT

A 30-year-old married female came to us with a slow growing mass in the right side of perineum since four years. Symptoms were mild in the form of dull aching pain, difficulty during defecation and sexual intercourse. On examination a large globular, non-tender, firm mass, bulging from right side of perineum was present. The per-rectal digital examination revealed the obliterated anorectal canal, mass felt in the right ischio-rectal fossa, pushing the wall to the left. The wall was not fixed to the mass. Similarly, vagina was partially obliterated and deviated to left. There was no local lymphadenopathy or dilated veins. Contrast enhanced computed tomography (CECT) scan was done, showing a heterogeneous soft tissue mass of 9´9´10 cm size in the right ischio-rectal region and pelvis, pushing anorectum, vagina and cervix on left side and gluteal muscle posteriorly, with no infiltration into the surrounding structures (Figure 1). On the basis of clinical findings and CT scan a soft tissue sarcoma was suspected. The routine haematological and biochemical tests were within normal limit. Fine needle aspiration cytology (FNAC) was, expectedly, inconclusive. Definitive surgery was planned. The patient was placed in jack knife position. Elliptical incision of 10

cm size was given across the bulge, further making it cruciate for adequate access. The mass was retrieved with an intact fibrous tissue capsule from the right ischio-rectal fossa after combined blunt and sharp dissection. Important anatomical structures like the anal canal, anal sphincter muscles, perineal body, sciatic nerve, pudendal and inferior rectal vessels and nerves were identified and preserved. Primary closure of surgical wound was done with a suction drain in the cavity. The postoperative recovery was uneventful and the drain was removed on the fifth post-operative day.

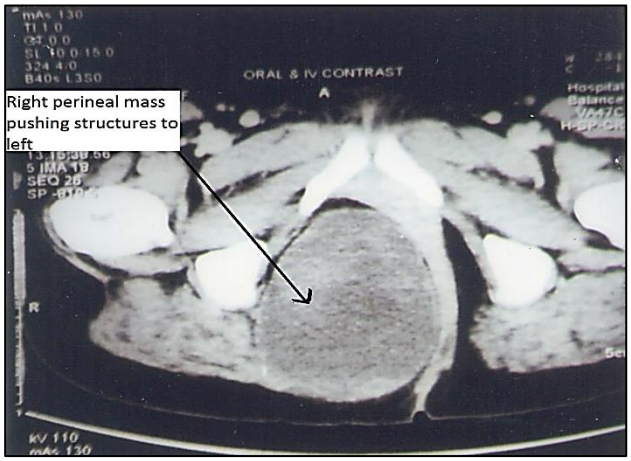

Figure 1: A section of CT scan shows mass in the right perineum pushing the perineal structures to the left.

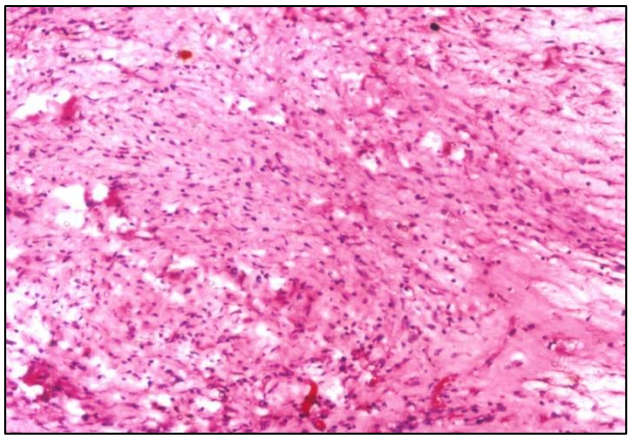

Figure 2: Low grade fibromyxoid sarcoma, photomicrograph showing mildly atypical spindled/oval cells disposed in a fibromyxoid stroma. There is confluence of myxoid zones seen in the right of the micrograph. Haematoxylin and Eosin X 40.

On gross appearance, the mass was firm, surrounded by a fibrous capsule with bosselated surface. The cut section showed greyish white appearance with patchy areas of small haemorrhages. The histopathology of the mass came out to be low-grade fibromyxoid sarcoma (LGFMS) (Figure 2). It was further found to show strongly positive immunoreaction to vimentin. The patient is still in follow up for last 5 years with no features of recurrence. Patient has preserved anal continence with a satisfactory sexual life but occasional shooting pain locally.

DISCUSSION

The term low grade fibromyxoid sarcoma (LGFMS) was first coined by Evans in 1987.[1] It is a rare, indolent, but potentially metastasizing tumour, belonging to fibroblastic/myofibroblastic soft tissue tumour category.[2,3] An incidence of 0.18 per million has been reported.[4] It affects mainly young to middle-aged adults. However, it is increasingly being reported in children and elderly patients.[5] The sex ratio is almost equal with slight male dominance, however Maretty-Nielsen et al. reported a female predominance.[2,4,6] This tumour is characterized by a painless, slow growing mass particularly in proximal lower extremities especially thigh.[2]

Majority of LGFMS occur in sub-fascial locations arising from deep subcutaneous tissue and skeletal muscles, but the involvement of superficial locations like sub-cutis or dermis are also increasingly being reported.[2-5] It has now been extensively reported in most parts of body including many intra-abdominal locations, shoulder, chest wall, axilla, inguinal region, vulva, buttocks, neck, mediastinum, brain and perineum.[2-8] Magnetic resonance imaging (MRI) is the investigation of choice, however, in our case CT scan was done as MRI was not available at our center.[4] Positron emission tomography–computed tomography (PET-CT) can also be used but is reported to be inferior to MRI.[4]

The fibrous content is typically iso-dense, and myxoid content hypodense, to muscle on non-contrast CT scan. In MRI, fibrous component is seen to be hypointense on T1 and T2-weighted images while myxoid component is hypointense on T1 and hyperintense on T1-weighted images. On gadolinium enhanced MRI, myxoid areas show intense enhancement while fibrous component only enhances slightly on T1-wighted images.[9] Imaging in LGFMS, while helpful in experienced hands, is still highly non-specific. The needle aspirates are hypo cellular with an abundant myxoid background so don't contribute much to diagnosis. Grossly, LGFMS appears as a well circumscribed, oval to round mass with a thin, fibrous pseudo-capsule.[1,2,6] The cut surface shows whorled, white-gray, firm and fibrous consistency with homogenous appearance.

The typical histological picture of LGFMS comprises of two components, fibrous and spindle cell with linear arrangement, producing whorled and swirling growth pattern, myxoid areas with spindle to stellate shaped cells with abundant intercellular matrix.[1,2] Folpe et al. have also found in some cases, areas of hypercellularity, nuclear enlargement and hyperchromatism, indicating presence of focal area of intermediate and high grade sarcoma.[6] The IHC of this tumour showed diffuse and strong reactivity for vimentin but are generally negative

for cytokeratin, smooth muscle actin, S-100 protein and neuron specific enolase.[2,7] The chimeric FUS/CREB3L2 gene detected by reverse transcription polymerase chain reaction (RT-PCR) assay or fluorescent in situ hybridization (FISH) technique seems to be specific for LGFMS and its expression in the t(7;16) (q33p11) is a useful tool for the diagnosis.[2,8,10] The cytogenetic analysis also reveals that low-grade fibromyxoid sarcoma and hyalinizing spindle cell tumour with giant rosettes are variants of the same entity having a characteristic chromosomal abnormality, t (7;16), (q34;p11).[11]

The diagnosis is made on histopathology, IHC and cytogenetic evaluation. Differential diagnosis in this condition includes desmoid fibromatosis, aggressive angiomyxoma, sclerosing epithelioid fibrosarcoma, soft tissue perineurioma, intramuscular myxoma, myxoid liposarcoma, myxoid variant of dermatofibrosarcoma protuberans and low grade myxofibrosarcoma. Wide local surgical excision is the only mode of treatment.[4] Not much has been reported and discussed about the details of surgical management of this condition. Most of these tumours are deceptively well-circumscribed but not encapsulated, though our tumor was well encapsulated.[2]

This makes the resection, often, incomplete; and thus leading to the commonly reported local recurrences and also contributing to their late metastasizing nature.[2] Meticulous dissection with preservation of vital perineal structures is, thus, essential for providing such young patients an optimal quality of life. De-differentiation is a reported feature which leads to early local recurrence and metastasis leading to a shorter survival in this, otherwise, long-survival-reported disease.[8] Radiotherapy and chemotherapy (with special reference to trabectedin) have also been tried but with little success; and thus only reserved for unresectable, recurrent and metastatic cases.[4]

The local recurrence occurs in approximately 10% cases and distant metastasis occurs in 5-10% cases.[6,8] Lungs appear to be the most common site of metastasis. Late metastasis is a well-documented and characteristic feature of LGMFS, with metastasis occurring as late as up to 45 years.[2,8] Thus, once diagnosed, LGMFS requires lifelong surveillance.[2,4,8] Depending upon location, patient can be followed up clinically and/or radiologically using x-ray, ultrasonography, CT-scan or MRI.

CONCLUSION

Low-grade fibromyxoid sarcoma is a rare pathological entity and its presence in perineal area is exceedingly rare. Histopathological examination and IHC staining with vimentin revealed the diagnosis. Adequate surgical excision, though technically demanding, was possible without damaging the other vital structure. Thus, wide local excision with meticulous dissection is essential for preserving vital structures, preventing local recurrences and late metastasis, and providing an optimal quality of life to such young patients. No adjuvant therapy was considered despite of large tumour size, since adjuvant therapy has no reported role. In 5 years of clinical follow-up, there has been no recurrence.

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