Case Report

DOI: http://dx.doi.org/10.18203/2349-2902.isj20170880

Malignant peripheral nerve sheath tumor with lymph node metastasis: an uncommon presentation

Suruchi Shreshtha*

Department of Surgery, Dr Baba Saheb Ambedkar Medical College and Hospital, Delhi, India

Received: 05 January 2017 **Accepted:** 02 February 2017

*Correspondence: Dr. Suruchi Shreshtha,

E-mail: suruchishreshtha@yahoo.com

Copyright: © the author(s), publisher and licensee Medip Academy. This is an open-access article distributed under the terms of the Creative Commons Attribution Non-Commercial License, which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

MPNST is a rare soft tissue neoplasm that accounts for less than 10 % of all soft tissue sarcomas. These tumors often affect males between 20 to 50 years, often with NF1, These with predilection for buttocks, thigh, brachial plexus, upper arm and paraspinal region. MPNST spread perineurally or hematogenously to lungs. Lymph node metastasis is seen in less than 5% cases of all sarcomas. This is a case of 46 year old male with NF-1 with MPNST posterior abdominal wall. The patient had metastatic pleural effusion in the absence of lung metastasis, along with widespread axillary, retroperitoneal and inguinal lymph node metastasis. Thus, although lymph node metastasis from MPNST is rare, this case expands our knowledge of the clinical behavior of MPNST.

Keywords: Lymph node metastasis, Malignant peripheral nerve sheath tumor, S-100

INTRODUCTION

MPNST is a rare soft tissue neoplasm that accounts for less than 10 % of all soft tissue sarcomas. These tumors have a known association with neurofibromatosis type1 syndrome, an autosomal dominant disorder that involves NF-1 tumor suppressor gene, located on chromosome 17. These tumors often originate in pre-existing neurofibromatosis type 1 lesions, affecting males between 20 to 50 years of age. These tumors have predilection for buttocks, thigh, brachial plexus, upper arm and paraspinal region. MPNST spread perineurally by direct invasion or hematogenously to lungs followed by bone and finally pleura.

Lymph node metastasis is seen in less than 5% cases of all sarcomas and are often associated with epithelioid sarcoma, synovial sarcoma, rhabdomyosarcoma, clearcell sarcoma, and angiosarcoma. Lymph node metastasis is rare in MPNST and occurs in presence of widespread metastasis.³ MPNST is treated by surgical excision and lymph node dissection is not routinely performed.

CASE REPORT

A 46-year-old male patient presented with progressively increasing, painful, left flank swelling of one year duration. He also complained of swellings in left axilla and groin for last six months. There was a steady unintentional significant weight loss but no jaundice, fever, bowel or urinary complaints. Patient had history of smaller swelling over same site which was excised two years back and diagnosed as neurofibroma on histological examination.

On examination, the patient weighted 60 kilos had multiple neurofibromas (Figure 1) all over his body along with café au lait spots and axillary freckling. He also had left axillary and inguinal lymphadenopathy. Mild pitting edema over the ankles and feet was recorded. A solitary, 15x12 cm, firm, bosselated, non-tender parietal mass extending between left costal margin and left iliac crest, from left anterior axillary line to left posterior scapular line was palpable, with overlying scar of previous surgery (Figure 2). The mass had restricted mobility in all the

planes and was also fixed to the skin in the region of the scar. No free fluid could be demonstrable. Liver, spleen and kidneys were not palpable. External genitalia were normal and systemic examination revealed left pleural effusion. Per-rectal examination was non-contributory.



Figure 1: Patient with neurofibromatosis type I with multiple neurofibromas and café au lait spots.



Figure 2: Patient with neurofibromatosis type I with malignant nerve sheath tumor left flank.

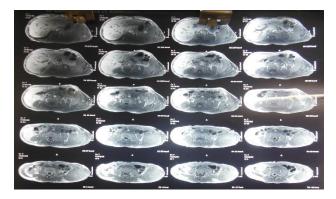


Figure 3: MRI abdomen of patient with lobulated subcutaneous soft tissue mass arising from posterolateral abdominal wall with retroperitoneal lymphadenopathy.

On investigation, a complete haemogram and urinalysis, biochemical investigations including urea, creatinine, sugar and liver function tests were within the normal range. A radiograph of the chest revealed left pleural effusion, which was positive for malignant cells suggestive of adenocarcinoma. Ophthalmic examination revealed bilateral corneal nevi and lisch nodules over iris. Incisional and lymph node biopsy were suggestive of epitheloid variant of malignant peripheral nerve sheath tumour with lymph node metastasis, and stained positive for vimentin and S-100. The tumor tissue stained negative for HMB-45, CK, EMA, Melan- A, DesminSMA and CD34.

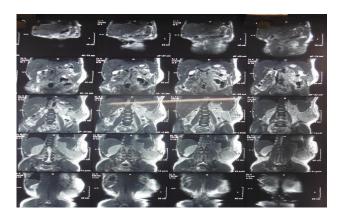


Figure 4: Coronal sections of the same MRI abdomen showing lobulated subcutaneous soft tissue mass arising from posterolateral abdominal wall with retroperitoneal lymphadenopathy and left pleural effusion.

abdomen **MRI** revealed single 9.4x8.5x3cm multilobulated subcutaneous soft tissue mass suggestive of sarcoma, arising from left posterolateral abdominal wall muscles with necrotic areas (Figure 3 and 4). There was also retroperitoneal lymphadenopathy in preaortic and para aortic regions, hepatomegaly and cholelithiasis without liver metastasis. There were multiple subcutaneous swellings. Other organs were normal. CECT chest revealed left pleural effusion but no lung metastasis. Bone scan also did not reveal any bony metastasis.

The patient was taken up for chemotherapy with alternating cycles of cyclophosphamide/doxorubicin and cisplatin/etoposide in view of metastatic tumor. Patient is doing well so far, pending further follow-up.

DISCUSSION

MPNST is a rare neoplasm accounting for less than 10% of all soft tissue sarcomas with an incidence of 0.1/100,000 per year. A sarcoma is defined as MPNST when at least one of the following criteria is met:

- It arises from a peripheral nerve
- It arises from a preexisting benign nerve sheath tumor (neurofibroma)
- It demonstrates schwannoma cell differentiation on histologic examination.

These tumors have a known association with neurofibromatosis type 1 syndrome, an autosomal dominant disorder that involves NF-1 tumor suppressor gene, located on chromosome 17. It affects individuals between 20 to 50 years of age and can grow to giant sizes. These tumors often affect males, and often involving buttock, thigh, brachial plexus, upper arm and paraspinal region. Incidence of MPNST in patients with NF-1 is not exactly clear, varying from 2-29% in different literatures.2 MPNST has been found to be associated with p53, p16 mutations, while NF1 gene activity does not independently cause MPNST. It can arise de-novo however; up to 60% MPNST originate in pre-existing neurofibromatosis type 1 lesions. Nerve of origin is not identifiable in upto 61% cases as documented by Nambisan et al.4 The tumor spreads perineurally by direct invasion or hematogenously to lungs followed by bone and finally pleura. Definitive diagnosis requires tissue specimen in the form of FNAC, trucut biopsy or incisional biopsy, latter is recommended. Immunohistochemistry and electron microscopic studies are gradually replacing histopathology as confirmatory diagnostic methods, as these tumors display a wide variety of histological patterns and hence no absolute standard is accepted.⁵ Studies by Matsunou et al have identified S-100 protein positivity and cytokeratin negativity as a supplementary tool for confirmation. The tumor is found to be S-100 positive in approximately 50-90% on immunohistochemistry. The treatment for MPNST has been combined modality that includes aggressive surgical excision or mass debulking with several cycles of adjuvant chemotherapy with cyclophosphamide/doxorubicin have been advocated. Adjuvant radiotherapy has also been investigated for treatment of advanced MPNST. For metastatic tumors chemotherapy is used as palliative therapy. Lymph node metastasis is rarely seen in patients with MPNST.3 Very few cases exist in the literature that demonstrates lymph node metastasis in MPNST.⁷

Funding: No funding sources Conflict of interest: None declared Ethical approval: Not required

REFERENCES

- 1. Enzinger FM, Weiss SW. Malignant tumours of peripheral nerves. In: Enzinger FM, Weiss SW, eds. Soft tissue tumors. St. Louis: CV Mosby Company; 2001;31:1209-1263.
- 2. Sorensen SA, Mulvihill JJ, Nielsen A. Long-term follow up of von Recklinghausen neurofibromatosis. N Engl J Med. 1986;314:1010-15.
- 3. Cameron RB, Loehrer PJ, Thomas CR Jr. Neoplasms of the mediastinum. In: DeVita VT Jr, Hellman S, Rosenberg SA, eds. Cancer principles and practice of oncology. 6th ed. Lippincott Williams and Wilkins, USA; 2001:1031-2.
- 4. Nambisan RN, Rao U, Moore R. Malignant soft tissue tumors of nerve sheath origin. J Surg Oncol. 1984;25:268-72.
- 5. Ducatman BS, Scheithauer BW, Piepgras DG. Malignant peripheral nerve sheath tumor: A clinicopathological study of 120 cases. Cancer. 1980;87:2006-21.
- Matsunou H, Shimada T, Kakimotos S. Histopathological and immunohistochemical study of malignant tumor of peripheral nerve sheath (malignant schwannoma). Cancer. 1985;56:2269-79.
- 7. Strauss M, Grey L. Malignant tumors of the peripheral nerves. In: Weiss SW, Goldblum JR, eds. Enzinger and Weiss's soft tissue tumors. 4th ed. St. Louis: Mosby; 2001:903-944.

Cite this article as: Shreshtha S. Malignant peripheral nerve sheath tumor with lymph node metastasis: an uncommon presentation. Int Surg J 2017;4:1138-40.