MALIGNANT PERIPHERAL NERVE SHEATH TUMOURS: A DIAGNOSTIC DILEMMA

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Abstract: Malignant peripheral nerve sheath tumor is a rare and aggressive type of soft tissue tumors and accounts for approximately 5-10% in soft tissue sarcomas.[1]. Mostly these are associated with Neurofibromatosis type-1. Here is a case of 35 year old woman having mass over left infraclavicular region. She undergone complete resection of tumor with adjuvant radiotherapy. She has been monitored and followed up since 4 months without any signs of local recurrence.

Keywords: Malignant nerve sheath tumor, Neurofibromatosis, Radiotherapy, Histomorphology

INTRODUCTION:

Malignant peripheral nerve sheath tumors (MPNST) usually occurs along the course of myelinated nerves, but they commonly appear in or near a nerve of the trunk or the limbs. In 50% of cases, they are associated with neurofibromatosis type 1(NF1). 40–47% develop sporadically and rest may develop post-radiotherapy.[2]. The most frequent sites of MPNST metastasis are lung, liver, brain, soft tissue, bone, regional lymph nodes, and retro-peritoneum. These tumours are most of the time painless but can produce compressive symptoms such as motor and sensory dysfunction. They are associated with a high risk of local recurrence ranging from 32% to 65% and hematogenous metastasis [3]. The sensitive markers involved consist of S-100 and SOX10 [4]. The diagnosis of MPNST in a patient without diagnosed neurofibromatosis needs to be evaluated carefully on the basis of the presence of the spindle cell sarcoma markers [5]. The aim of this paper to report a case of MPNST in clinically nonestablished NF-1. The pattern of occurrence and histomorphological diversity leads to both diagnostic and management dilemmas.

CASE

A 35 year old female presented to surgical opd with complaints of swelling over left side of axilla since 6 months which was gradually and progressively increasing in size since 4 months but since last 1 month it is increasing rapidly. It is not having any complaints of pain, sensory or motor dysfunction but swelling affects her daily routine.

On examination -approximately 6x4cm² size illdefined diffuse swelling felt in left infraclavicular region which has firm consistency. No local rise of temperature or tenderness felt.
On ultrasonography-57x33mm size of well-defined, heterogeneous, hypoechoic lesion with few echogenic foci (calcification) and internal vascularity noted in intramuscular plane in left infraclavicular region. Neoplastic etiology likely.

In Contrast CT Neck And Thorax we got 61x60x43mm heterogeneously enhancing lesion involving left pectoralis minor muscle. Lesion is abutting left subclavian and axillary artery. Lesion is also encasing left left axillary artery and abutting left pectoralis major muscle. No bony erosion or intrathoracic extension noted.

Complete excision of tumor mass has been done. According do radiation oncologist, radiotherapy has been started and since then regular followup of patient has been taken with no signs of local recurrence since 4 months.

**DISCUSSION:**

MPNST are tumors which originates from schwann cells or fibroblast of peripheral nerve sheaths. They can arise from all nerves but more commonly from large nerves and trunks of sciatic nerve and brachial plexus. MPNST falls into soft tissue sarcomas but very uncommon although they are more common in association with neurofibromatosis type -1 but very rare in general population. MPNST presents as mass which is firm in consistency which is rapidly increasing in size over time. Most of them are painless to begin with but soon have compression symptoms such as radiculopathy, sensory and motor deficits. Patient with Neurofibromatosis may misdiagnose with it a plexiform neurofibroma and presents relatively late than a general population who reaches soon with a complaints on firm swelling. MPNST share many similarities with other benign soft tissue tumors so it is not simple to reach an accurate diagnosis of it even with help of radiology. MRI is diagnosis of choice. MRI shows intratumoral cystic masses with peripheral enhancement patterns. Wide tumor excision with appropriate tumor free margin is treatment of choice. In case where sufficient margin cannot be obtained adjuvant radiotherapy should be given to prevent local recurrences. The lungs, liver, bones are most common sites for local recurrence and metastases in MPNST. Mpnst poses challenge for pathologist to diagnose it as it shares many histomorphologic similarities with other soft tissue tumors and adequate immunologic markers and FISH technology required for appropriate diagnosis. Cellular schwannoma, leiomyosarcoma, MPNST, synovial sarcoma, and clear cell sarcoma are among the differential diagnoses for soft-tissue spindle cell sarcomas. S-100 and NSE are specific tumor markers for MPNST. SMA, vimentin and desmin are for leiomyosarcoma. S-100, epithelial membrane antigen
(EMA), and vimentin for Schwannoma, cytokeratin and vimentin for synovial sarcoma, vimentin for fibrosarcoma, and S-100, melanin, and glycogen for clear cell sarcoma. We cannot differentiate without these markers among these high grade tumors. MPNST and synovial sarcoma further needs FISH to exclude SYT–SSX gene fusion for final confirmation.

Conclusion:
The doctor-patient relationship is emphasized in such cases and emotional commitment of the patient towards the surgeons are to be stressed for the new generation clinicians. Such cases are challenging to diagnose and accurate management is also of utmost importance. This case can help on how to approach, make differential diagnosis and reach the final one and also emphasizes on aggressive management in such tumors as they can improve mortality and morbidity in patients.

REFERENCES:
1. R.E. Ferner, D.H. Gutmann
   International consensus statement on malignant peripheral nerve sheath tumors in neurofibromatosis
   View in Scopus Google Scholar
5. A.C. Gamboa, A. Gronchi, K. Cardona
   Soft-tissue sarcoma in adults: an update on the current state of histotype-specific management in an era of personalized medicine