Malignant Peripheral Nerve Sheath Tumour Presenting with Horner’s Syndrome

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Abstract

A young male presented with clinical and radiological features of right apical lung mass and Horner’s syndrome. Subsequently the patient was diagnosed as a case of malignant peripheral nerve sheath tumour (MPNST) at the apex of right lung originating from an intercostal nerve and compressing ipsilateral cervical sympathetic plexus and lower cord of brachial plexus, in a case of neurofibromatosis type 1.

Introduction

In the Department of Pulmonary Medicine, apical bronchogenic carcinoma is the commonest cause of Horner syndrome with brachial plexopathy. We are reporting a case of malignant peripheral nerve sheath tumour in a case of neurofibromatosis type 1, who presented with Horner’s syndrome with brachial plexopathy. This is a very rare presentation of its kind, only a small number of such cases have been published so far.

Case History

A 23 year old male, non diabetic and normotensive, mason by occupation, presented with dull aching pain in right shoulder and anterior chest wall for 1 year which radiated to inner aspect of arm and fore arm. He also complained of weakness of right hand, loss of sweating of right side of face and shrunken right eye for 1 year.

Examination: The patient had mild pallor, multiple neurofibromas on skin, cafe-au-lait macules > 6 in number and each >1.5cm in diameter, axillary and inguinal freckles, features of Horner’s syndrome and lisch nodules on slit lamp examination of eyes. Chest examination revealed decreased movement of right upper chest wall, dull percussion note in right supraclavicular and suprascapular areas, 2nd and 3rd intercostal spaces in right mid clavicular line and diminished vesicular breath sound in corresponding areas. On neurological examination, loss of fine touch and pin prick sensation on inner aspect of right forearm and hand, atrophy of hypothenar eminence and weakness of 3rd and 4th lumbricals were found. The neurological features were diagnostic of Horner’s syndrome with brachial plexopathy (lower cord).

Investigations: NCV examination was suggestive of C8, T1 involvement. CXR showed a right apical lung mass. CT-scan of thorax showed a well defined irregular, enhancing mass in right upper lobe (Figure 1). CT-guided FNAC from the lesion revealed scanty cellularity with few spindle cells. The patient was subjected to thoracotomy and resection of entire tumour mass of about 8.5 cm diameter (Figure 2), which was found to arise from apex of right posterior chest wall, was done. Histopathological examination reported it to be a case of peripheral malignant nerve sheath tumour, intermediate grade spindle cell sarcoma of undetermined histogenesis (Figure 3) without coagulative tumour cell necrosis. Immunohistochemistry study report showed that the tumour cells were immunonegative for Desmin, SMA, h-Caldesmon, CD 34 and focal positive for S-100 protein.

Management: The patient received chemotherapy with Vincristin, Cylcophosphamide and Doxorubicin followed by post operative radiotherapy.

Follow up: The patient is still under follow up. Features of Horner’s syndrome with right brachial plexopathy have partially improved.

Discussion

Horner’s syndrome is a triad of partial ptosis, meiosis and ipsilateral hemi facial anhidrosis

Neurofibromatosis type 1 is diagnosed by any two of the following criteria;

1. Multiple cafe-au-lait macules larger than 1.5 cm in diameter and more than six in number.
2. Axillary or inguinal freckles.
3. Two or more neurofibromas of any type or one plexiform neurofibroma.
4. Sphenoid wing dysplasia or congenital bowing of long bone cortex with or without pseudoarthrosis.
5. Bilateral optic nerve glioma, two or more Lisch nodules on slit lamp examination.
6. First degree relative with NF1 diagnosed by preceding criteria.

A sarcoma is defined as malignant peripheral nerve sheath tumour (MPNST) if at least two of the following criteria are met with.

It arises from a peripheral nerve.
It arises from a pre-existing benign nerve sheath tumour (neurofibroma).
It demonstrates Schwann cell differentiation on histological examination.

The incidence of malignant peripheral nerve sheath tumour (MPNST) in general population is 0.001%. Its incidence in patients with Neurofibromatosis type 1 (NF1) is 4.6%. Up to 50% of MPNST occurring in patients with NF1 arise from a pre-existing neurofibroma. NF1 patients have a 10% lifetime risk.
of ultimately developing MPNST. MPNST generally occurs in adulthood, typically between 20 and 50 years of age.\(^1\) It usually presents as an enlarging palpable mass. Pain is a variable complaint. Rapid enlargement occurs more often in patients of NF1. MPNST arising from a peripheral nerve may result in variable clinical patterns including radicular pain, paraesthesia and motor weakness of muscles. Most MPNST occur in conjunction with large peripheral nerves such as sciatic nerve, brachial plexus and sacral plexus. Haematogenous metastasis from MPNST occurs most commonly in lungs.\(^2\) The reported five year survival rate for patients with MPNST without NF1 is as high as 50%. It drops down to as low as 10% in MPNST patients with NF1.\(^2\)

Treatment consists of complete and wide surgical resection. Adjuvant irradiation seems to improve local control of the disease. Chemotherapy is only employed in high grade malignant disease in which metastasis is likely. Chemotherapy can be administered preoperatively or postoperatively. The prognosis depends on tumour size, surgical excision margin, and histological presence of necrosis and history of prior irradiation.\(^3\)

MPNST frequently recurs locally and metastasises distally with lung being the most common site. Malignancy has been reported to be the cause of about 25% of cases of preganglionic Horner’s syndrome with the most common tumour being lung and breast cancer.\(^4\) In our case, Horner’s syndrome was preganglionic in origin. MPNST in a case of NF1 is a very rare cause of Horner’s syndrome associated with brachial plexopathy. S-100 is traditionally regarded as the best marker for malignant peripheral nerve sheath tumour; however, it is positive in only about one third to half the tumours.\(^5\) The negative reaction may be caused by not only tumour cell anaplasia but also by different constituents other than Schwann cell. All perineural MPNST were S100 protein negative and EMA and/or GLUT1 positive. For the immuno histochemical diagnosis of MPNST, nerve sheath markers other than S100 protein such as CD57 (Leu7), EMA, Glut1, CD34 should also be included in the panel of antibodies.\(^6\) To the best of our knowledge, this is the first case reported online from India.

References