

Rare Presentation of Intrathoracic Nerve Sheath Tumour and Cervical Tumour in a Patient with Von Recklinghausen's Disease

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Abstract

An 11-years-old male presented with the history of inability to abduct right upper limb and weakness of bilateral upper limb (right > left) for one month. Clinically he was diagnosed to have Von Recklinghausen's disease based on National Institute of health (NIH) Consensus Development Conference Criteria. Magnetic Resonance Imaging (MRI) was suggestive of cervical tumour at C3-5 level and D2-6 level posterior mediastinal mass. He underwent excision of cervical tumour and laminotomy at C3-C5 level by the team of Neurosurgeons followed by posterior mediastinal mass excision by Thoracic Surgeons under General Anesthesia with double lumen tube intubation. This case emphasizes the rare presentation of cervical and intrathoracic posterior mediastinum nerve sheath tumour in a patient with Von Recklinghausen's Disease.

Keywords: National Institute of Health (NIH); Magnetic Resonance Imaging (MRI); Cervical Tumour; Von Recklinghausen's Disease

Introduction

Neurogenic tumours are the most common mediastinal tumours, accounting for 12% to 21% of all mediastinal neoplasms [1] and 75% to 95% of all posterior mediastinal neoplasms [2]. Intrathoracic neurogenic tumours usually originate from the intrathoracic nerve sheath, autonomic nerve ganglion sections and sub-organizations. Schwannoma and neurofibroma are the most common neurogenic tumour. Nearly half of posterior neurogenic tumours are asymptomatic, however, as they become larger in size, they can produce symptoms related to local compression, bone invasion, and spinal cord involvement [3]. Surgical excision is the treatment of choice.

Case Report

A 11 year old male presented with the history of inability to abduct right upper limb and weakness of the bilateral upper limb (Right > left) for one month. Patient had no respiratory complaints. Clinically patient had cafe au lait spots, axillary freckling and maternal history of diagnosed Neurofibromatosis type 1 (NF1). CECT thorax was suggestive of lobulated mildly enhancing heterogenous soft tissue density lesion in right paravertebral and paraspinal region extending from D2-D6 vertebral level. MRI cervical spine was suggestive of lobulated mass lesion at C3-C5 level extending into neural foramina. CT guided biopsy was inconclusive. He underwent surgical excision of cervical tumour with laminotomy of C3-C5 level by a team of Neurosurgeons followed by excision of posterior mediastinum mass by posterolateral thoracotomy. Postoperative outcome was uneventful and patient was able to abduct his right upper limb above the shoulder and weakness of bilateral upper limb subsided gradually. Histopathology was suggestive of cervical neurofibroma and thoracic plexiform neurofibroma.



Figure 1: Axillary freckling and Café au lait spots.



Figure 2: Chest X ray PA view.

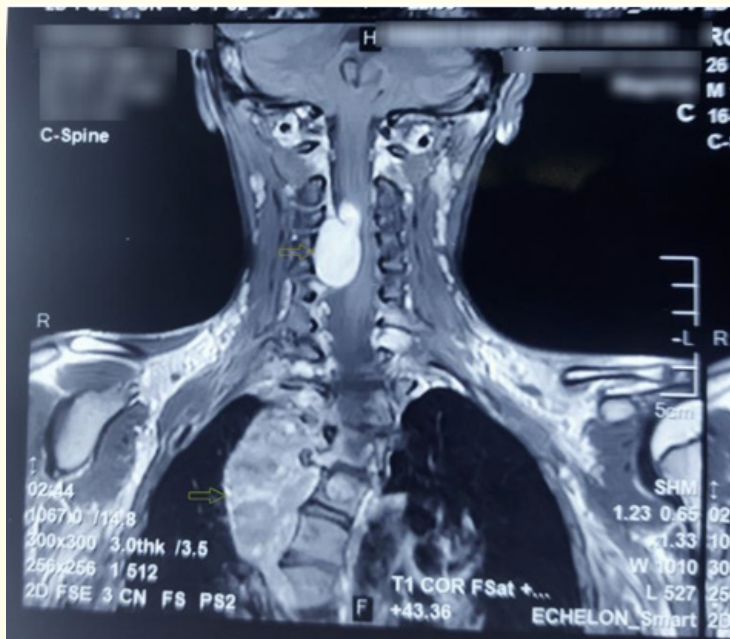


Figure 3: MRI cervico-thoracic spine.

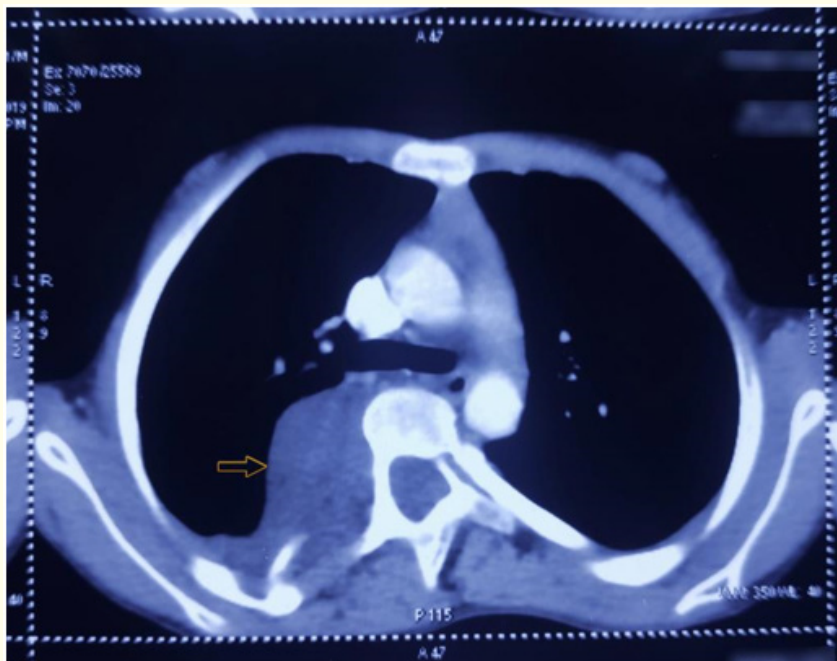


Figure 4: CT thorax.



Figure 5: Intraoperative image-posterior mediastinal mass.

Discussion

NF1 is an autosomal dominant neurocutaneous disorder. The NF1 gene mutation is on chromosome 17 and its product, neurofibromin, normally functions as a tumour suppressor to reduce cell proliferation by inactivating the proto-oncogene p21-ras [4]. NF1 patients, therefore, have an increased predilection for the development of both benign and malignant nerve sheath tumours. Mediastinal tumours in NF1 patients are typically neurofibromas that originate in the posterior mediastinum and have a particular 2 - 3% predisposition for malignant transformation [5].

Our patient was diagnosed to have Von Recklinghausen's disease as per below criteria. As per National Institutes of Health (NIH) consensus panel, diagnosis of NF1 is done as per the following. Patient should have two or more signs:

- Six or more café au lait macules larger than 5 mm in the greatest diameter in prepubertal children and larger than 1.5 cm in postpubertal individuals.
- Two or more neurofibromas of any type or one plexiform neurofibroma.
- Multiple freckles (Crowe sign) in the axillary or inguinal region.
- A distinctive osseous lesion such as sphenoid dysplasia or thinning of bone cortex with or without pseudoarthrosis.
- Optic glioma.
- Two or more iris hamatomas (Lisch nodules) on slit lamp examination.
- A first degree relative with NF1 as diagnosed by using above criteria.

CT scan usually provides sufficient information to assess the mediastinal mass; however, the radiologic features of benign and malignant peripheral nerve sheath tumours are nonspecific. MRI scan may further delineate intraspinal involvement and relationships to adjacent structures. The role for positron emission tomographic imaging remains undefined. The coexistence of retroperitoneal neurogenic tumours, although rare, should be recognized during the evaluation and management of NF1 patients with suspected mediastinal neurogenic tumours.

Excision of mediastinal neurogenic tumours is approached through a posterolateral thoracotomy. Thoracoscopic resection of neurogenic tumours is limited in patients with large (> 6 cm) tumour, suspected malignancy, intervertebral extension, and involvement of adjacent structures.

In our case patient presented with cervical mass with spinal canal extension at C3-C5 level and intrathoracic mass at D2-D6 level. He underwent surgical excision of cervical tumour with laminotomy of C3-C5 level by posterior midline incision in the neck by a team of Neurosurgeons followed by excision of posterior mediastinum mass by posterolateral thoracotomy. Entire tumour mass was removed en-bloc completely and sent for histopathological diagnosis. Postoperative outcome was fine and patient improved gradually.

Conclusion

This case emphasizes the rare presentation of cervical and intrathoracic posterior mediastinum nerve sheath tumour in a patient with Von Recklinghausen Disease and surgical approach with involvement at various levels and intraspinal extension.

Conflict of Interest

There is no conflict of interest.

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