PET avid lung mass in a neurofibromatosis patient: a case report

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Citation

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Abstract

The differential diagnosis of a lung mass in a neurofibromatosis patient includes benign or malignant neurogenic neoplasm. Benign neurogenic neoplasms can transform to malignant and since this transformation is a major cause of mortality, the prognosis will depend on early detection. Computed tomography scans, magnetic resonance imaging and positron emission tomography are not able to differentiate benign from malignant neurogenic neoplasm. Needle biopsy has poor sensitivity and malignant neurogenic neoplasm can be misinterpreted as benign. We address these issues in the case of a 48 year old man with neurofibromatosis type 1 who incidentally was found to have a right apex lung mass. The needle biopsy showed spindle cell lesion of probable neurogenic origin. Given the concern about the adequacy of needle biopsy in providing an accurate diagnosis, a positron emission tomography scan was done and it showed high uptake within the lesion. The mass was resected and confirmed as neurofibroma

INTRODUCTION

Neurofibromatosis is a disorder of neural crest cells such Schwann cells, melanocytes, and endoneurial fibroblasts. These cell types proliferate excessively throughout the body forming neoplasms(1). The neoplasms may be harmless or may cause serious damage by either compressing adjacent tissues or transforming into malignancy (2,3). Malignant neoplasms have a very high rate of local recurrence and tendency to metastasize. Therefore differentiating benign neurogenic neoplasms from malignant cases is crucial since early detection of malignancy improve the prognosis.

CASE PRESENTATION

A 48-year-old gentleman was recently admitted to the hospital with decompensated congestive heart failure (CHF). His echocardiogram showed diastolic dysfunction and moderately severe pulmonary hypertension. As part of his work up for his pulmonary hypertension a CT scan of the chest pulmonary embolus protocol was done which showed no evidence of pulmonary embolism, but a right apex pleural based mass measuring 2.7 cm. The patient was not aware of any prior history of lung mass. After treatment for his CHF, the patient only complained of a mild degree of dyspnea on excretion which was noted as a chronic symptom that had been stable for many years. The patient denied any chest pain or cough, fever or chills. His past medical history was significant for neurofibromatosis type 1 (NF-1), morbid obesity, hypertension, hyperlipidemia, obstructive sleep apnea syndrome, chronic obstructive pulmonary disease (COPD), allergic rhinitis, seizure disorder, depression, and chronic back pain. His medication list included Oxcarbazepine, Gabapentin, Atenolol, Lisinopril, Citalopram, Trazodone, Montelukast, Cetirizine, Tiotropium inhalation, and Ibuprofen. He has no known drug allergies. He smoked in the past but had quit many years ago. There was no history of alcohol or illicit drug abuse. His family history was significant for neurofibromatosis on his paternal side. His father and his paternal grandmother as well as his son have neurofibromatosis.

His physical exam was significant for multiple neurofibromatosis lesions, most pronounced on his left eyelid (Figure.1). There was one on left knee and multiple lesions over his anterior chest wall, his abdomen, and his neck. There were also multiple cafe au lait spots on his abdominal wall. Otherwise the rest of his physical exam was unremarkable.

He underwent CT guided transthoracic needle biopsy (figure 2) which showed spindle cell lesion of probable neurogenic origin. Given the concern about the adequacy of the needle biopsy in providing an accurate diagnosis, a FDG PET scan was ordered which showed high uptake within the lesion

with a Standard Uptake Value (SUV) of 7 (figure 3). The patient was referred for thoracic surgery and had an open thoracotomy revealing a mass arising from apical posterior region of the chest wall. The pathology was neurofibroma.

Figure 1

Figure 1: Picture showing neurofibromatosis lesions on the the left eyelid.

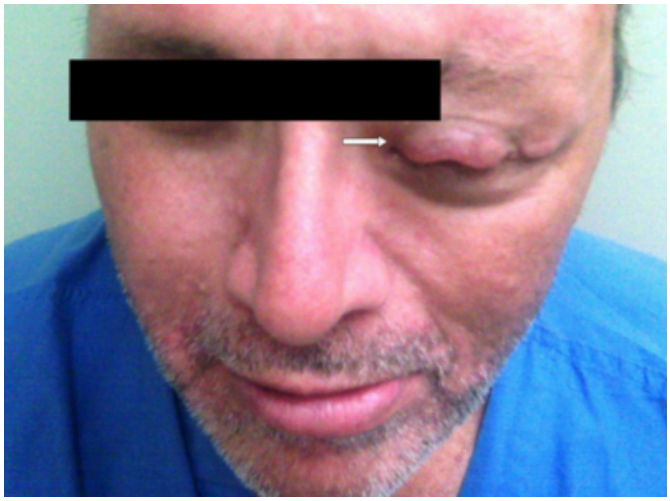


Figure 2

Figure 2: CT guided transthoracic needle biopsy from the lung mass

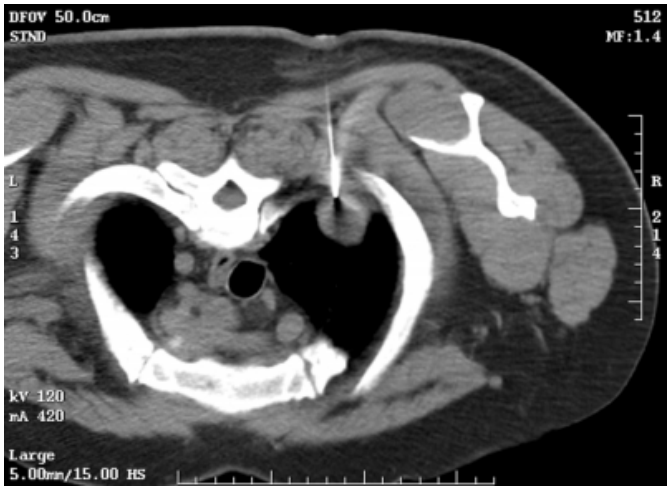
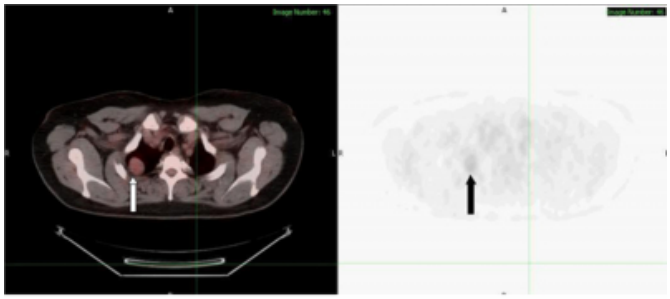


Figure 3

Figure 3: CT/PET scan showing high FDG uptake in the lung mass



DISCUSSION

A lung mass in a neurofibromatosis type 1(NF1) patient is not uncommon. Differential diagnosis of a lung mass in a neurofibromatosis patient will mainly include benign neurogenic neoplasm (BNN), and malignant neurgenic neoplasm (MNN). BNNs have the capacity to transform to malignant ones and this transformation is the major cause of mortality in neurofibromatosis patients. Malignant neoplasms have a very high rate of local recurrence and propensity to metastasize. Therefore, the final prognosis will depend on early diagnosis (1, 2).

Clinical symptoms and signs of MNNs include persistent pain and signs of local compression to adjacent structures such as bronchus or esophagus. However, these symptoms or signs may be seen in both benign and malignant lesions (3).

A Computed Tomography (CT) scan and Magnetic Resonance Imaging (MRI) can identify some malignant features such as a large infiltrative mass with irregular border and necrotic and heterogeneous contrast enhancement. These radiographic features are nonspecific which makes the CT scan and MRI unreliable in accurately characterizing a lesion as a benign or malignant (4,5,6).

Fluorodeoxyglucose (FDG) Positron Emission Tomography (PET) is a very sensitive tool in detecting MNNs in patients with NF1 (sensitivity=95%). However its specificity for MNNs in NF1 is only 70-80%. There is a significant overlap in the SUV measurement between benign and malignant neurogenic lesions with multiple BNNs having SUV values in the MNNs' range (7).

The sensitivity of the core needle biopsy in diagnosing BNNs is 60 %. Neurofibromatous areas of neurogenic sarcoma or low grade sarcoma may be misinterpreted as BNNs by percutaneous biopsy (8).

CONCLUSION

Neurogenic neoplasms are the main cause of intrathoracic masses in NF1 patients. Differentiating malignant and benign neurogenic tumors is highly important because early diagnosis of malignancy can improve prognosis. CT scans, MRI and PET are not specific alone for differentiating between benign and malignant neurogenic tumors but used in conjunction can improve the diagnosis. Needle biopsy alone has poor sensitivity and some MNNs can be misinterpreted as BNNs. Excisional surgical biopsy is the ultimate way to assure accurate diagnosis of PET avid lesions.

ABBREVIATIONS

BNN: Benign Neurogenic Neoplasm, MNN: Malignant Neurogenic Neoplasm, CT: Computed Tomography, MRI: Magnetic Resonance Imaging, PET: Positron Emission Tomography, NF1: Neurofibromatosis type 1, SUV: Standard Uptake Value, FDG: Fluorodeoxyglucose.

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