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Case Report

Rapidly Growing Giant Intrathoracic Neurofibroma with Massive Pleural Effusion

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Abstract

Neurofibromatosis type 1 is an autosomal dominant benign disease with variable expressions, usually involving nerves and skin. Intrathoracic rapidly spreading tumors in patients with Von Reclinghausen's disease is rare. Mainstay of treatment is the surgical resection of tumour in its early stage of diagnosis.

Keywords: Intrathoracic; Giant; Neurofibroma; Pleural Effusion

Introduction

Neurofibromatosis type 1 (NF-1) is an autosomal dominant benign disease with variable expressions, usually involving nerves (neurofibromas) and skin (pigmentation).NF-1 is doused by mutations in gene involving chromosome 17 with reported incidence of 1 in 4000 life births [1]. Intrathoracic tumour in patients of NF-1 have been reported in various case reports in the past, but intrathoracic giant rapidly growing benign neurofibroma with marked mediastinal shift and pleural effusion is rarely reported in the literature [2]. Patients with rapidly progressing thoracic NF-1 tumors should be adequately investigated to determine the possibility of surgical management. Here, we present the case of rapidly progressive giant intrathoracic neurofibromas in middle-aged women.

Case Presentation

We confronted a 35-year-old female presented to us in the emergency department with complaints of left side chest pain and shortness of breath for 3 months. On examination, multiple skin lesions (neurofibroma) and pigmentation of skin (café'-au-lait spots) and brown dome shaped lesions (neurofibromas) on the body were identified (Figure 1a). On respiratory examination trachea deviated to the right side and left side decreased breath sound. The infraclavicular and infra-mammary areas of the chest was a stony dull note on percussion. Chest x-ray showed complete white opacification of left lung with significant right sided mediastinal (Figure 1B). Chest contrast enhancement computed tomography CECT scan revealed ill defined heterogeneously enhancing mass lesions in left lingular segment, lower lobe, and right upper lobe with surrounding ground glass opacity GGO's and infiltration of pericardium and chest wall with mild left side pleural effusion (Figure 2). Ultrasound guided trucut biopsy taken from left lung mass and histology revealed benign neurofibroma, no significant mitotic figures were seen. Pleural fluid investigation revealed haemorrhagic, pleural fluid hematocrit level of 32% (more than 50% of blood hematocrit level), low (22 IU/L) adenosine deaminase (ADA), no malignant cells on cytology and culture was sterile. Pleural fluid for AFB (acid fast bacilli) and AFB culture came to be negative for tuberculosis. Post thoracentesis (1200 ml) blood hemoglobin dropped from 10.1 mg/dL to 8.5 mg/dL. In view of the massive hemothorax patient was planned for CT aortogram and cardiovascular surgery reference to evaluate the cause of hemothorax. Unfortunately the patient showed clinical and radiological worsening, before further work up for the cause of hemothorax and she died despite adequate medical management due to severe type 1 respiratory failure.

Discussion

The NF-1 gene is located on chromosome 17p11.2 and mutations involving these genes are responsible for the disease [3]. The increased risk of developing benign NF-1 tumours in the individuals is due to loss of neurofibromin. Our patients showed more than 6 café'-au-lait spots and multiple neurofibromas on chest and neck areas which are the classical characteristics features of NF-1. Other features of NF-1 are optic pathway glioma, pseudoarthrosis, freckling in axillary and groin [1,2]. Neurofibromas originate from fibroblast, Schwannand cells and any peripheral nerves. Hence, Von Reclinghausen's disease may involve the lungs, chest wall, ribs, and mediastinum. The possible differentials in cases of giant intrathoracic tumours include malignant peripheral nerve sheath tumours and intrathoracic meningocele. Mainstay of treatment in NF-1 is the surgical resection of tumour in its early stage of diagnosis and resection should be complete in one stage [4]. However, awareness of NF-1disease among the health care workers and genetic counseling should be recommended for patients and families. There are few case reports of massive spontaneous hemothorax in NF-1 patients. Hemothorax as a complication of benign neurofibroma is rare, and fetal especially when it is associated with rapidly growing intrathoracic lesions of NF-1, with associated mortality rate upto 28% [5]. As in our case it has been affected more often in females (63%) than males with more predilection to the left-sided predominance (61%). Mostly intercostal arteries (30%) and subclavian arteries (26%) are the source of bleeding, both of these arteries can be invaded by a neurofibroma [6]. Unfortunately in this case we could not be able to identify the source of bleed definitively due to the rapid progression of disease. The possible therapeutic approaches would be open surgery or interventional radiological procedures like intercostal artery embolisation or stenting [5,6]. Hemodynamically unstable patients should undergo thoracotomy to identify the cause of bleeding so that further suture ligation, resection of the neurofibroma or packing can be done to prevent bleeding.

Conclusion

Intrathoracic rapidly spreading tumors in patients with Von Reclinghausen's disease is rare but intrathoracic tumors can lead to rapid progression of disease with complications Loke hemothorax can lead to fatal outcomes if early diagnosis is missed. Hence, if a giant progressive thoracic NF-1 tumours with progressive effusion should be identified as soon as possible, and early surgical resection and surgery or interventional radiological procedures should be attempted to prevent the fatal consequences.

Bibliography

1. Ghalayani P., *et al*. "Neurofibromatosis type I (von Recklinghausen's disease): A family case report and literature review". *Dental Research Journal (Isfahan)* 9.4 (2012): 483-488.

2. Kesieme EB., *et al*. "Late presentation of giant intrathoracic neurofibroma with significant mediastinal shift: a case report and review of the literature". *Case Reports in Pulmonology* 20 (2013): 619729.

3. Friedman JM. "Neurofibromatosis 1". 1998 Oct 2 [Updated 2019 Jun 6]. In: Adam MP, Ardinger HH, Pagon RA, *et al*., editors. GeneReviews®. Seattle (WA): University of Washington, Seattle; 1993-2021.

4. Guo R., *et al*. "Giant atypical Neurofibroma in the right thoracic cavity of a 57-year old man: a case report with review of the literature". *North American Journal of Medical Sciences* 2 (2009): 135-136.

5. Hartzell HC. "Spontaneous Hemopneumothorax: Report of Three Cases and Review of Literature". *Annals of Internal Medicine* 17 (1942): 496.

6. Zacarias Föhrding L., *et al*. "A case of lethal spontaneous massive hemothorax in a patient with neurofibromatosis 1". *Journal of Cardiothoracic Surgery* 9 (2014): 172.