Case Report

Spontaneous hemothorax: A rare lethal pulmonary complication of neurofibromatosis type 1

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ABSTRACT

Neurofibromatosis 1 (NF1), also called von Recklinghausen's disease, is an autosomal dominant disease characterized by multiple non-cancerous tumors of nerves and skin, and areas of abnormal pigmentation. Vasculopathy and spontaneous hemothorax are rare complications, but potentially lethal, which necessitates quick and decisive intervention to save the life of the patient. Here, we present a case of spontaneous massive haemothorax leading to hemorrhagic shock in a 66-year-old woman with neurofibromatosis type-1. She was investigated and managed successfully.

Keywords: Neurofibromatosis type-1, Spontaneous hemothorax, Vasculopathy, Hemorrhagic shock

Neurofibromatosis 1 (NF1) is an autosomal dominant inherited disease caused by an abnormality of the long arm of chromosome 17 [1]. The disease is characterized by café-au-lait macules, neurofibromas, axillary and inguinal freckling, and Lisch nodules in the iris. Patients with NF1 may be associated with optic gliomas, distinctive osseous abnormalities, Schwann cell tumors, macrocephaly, intracranial tumors, kyphoscoliosis, syringomyelia, and pheochromocytoma. In addition, numerous vascular manifestations like vascular stenosis and aneurysms have been reported [2]. Spontaneous haemothorax is a rare but potentially lethal complication of these abnormalities [3].

CASE REPORT

A 66-year-old female presented to the emergency department with a complaint of difficulty in breathing for 5–6 h. There was no history of chest trauma. Her father was also suffering from neurofibromatosis and died at the same age due to an unexplained etiology.

On general physical examination, she was conscious, had tachypnea, and tachycardia, and the extremities were cold. Her blood pressure was 88/64 mm of Hg, heart rate was 124 beats/min, and respiratory rate was 34 breaths/min. Oxygen saturation at room air was 82%, grade 1 pallor in the general examination. In addition, she had multiple café-au-lait spots on her trunk, neurofibromas all over the body, and had severe cervical kyphoscoliosis (Fig. 1). Chest examination revealed evidence of absent breath sounds and decreased movement of the right side suggestive of some space-occupying lesion, like massive pleural effusion or mass lesion.

She was immediately resuscitated along the line of shock in the emergency by intravenous fluid, noradrenaline infusion, antibiotics, and other supportive measures. Simultaneously, other investigations were done, which revealed severe anemia with hemoglobin of 5.4 gm%, normal total leukocyte counts, and platelets.

Electrocardiogram, cardiac Troponins, 2D Echocardiography were normal. The chest X-ray suggests complete opacification of the right side of the hemothorax with a shift of the mediastinum to the left side. There was also evidence of scoliosis in the cervical area (Fig. 2). Ultrasound-guided fluid aspiration was done from the right side which suggested frank hemothorax. After aspiration of about 300 ml of blood, the patient became relatively comfortable and then, reinvestigation was planned. As the etiology was not clear, she underwent a Computed tomography (CT) scan of the thorax and CT pulmonary angiography to identify the site of bleeding, any mass lesion, and any vascular abnormalities. CT scan and angiography revealed significant fluid collection in the right hemothorax but no evidence of any mass lesion or active bleed (Fig. 3). There was a small blood clot in the upper part of the posterior mediastinum that could be the site of the bleed. All bleeding parameters were within normal limits. Aspirated fluid

Access this article online	
Received - 05 August 2021 Initial Review - 21 August 2021 Accepted - 22 September 2021	Quick Response code
DOI: 10.32677/ijcr.v7i10.3062	

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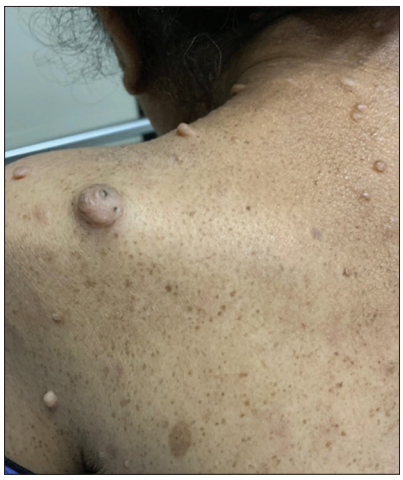

Figure 1: Multiple café-au-lait spots with neurofibromas

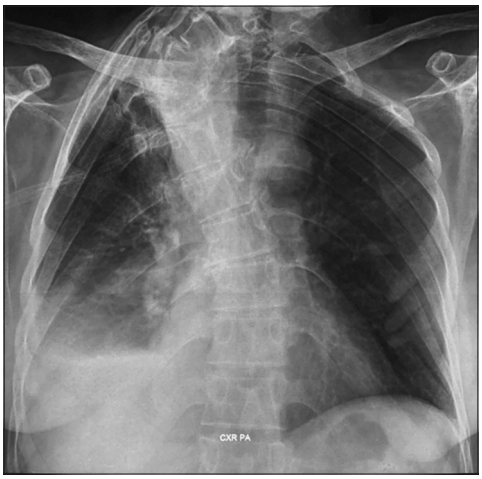

Figure 2: Chest X-ray revealed expanded lung on the right side, ribbon ribs with cervical scoliosis and pleural fibrosis

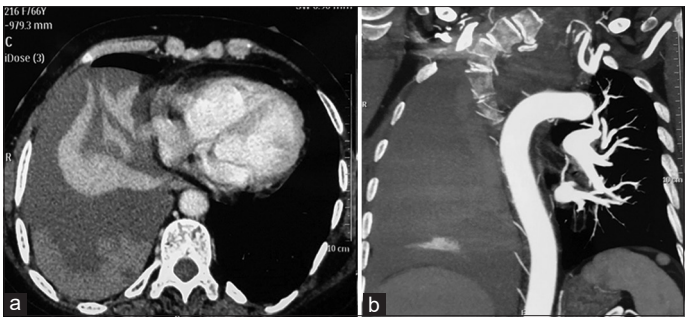

Figure 3: (a) CT scan thorax showing large hemothorax on the right side with blood clots, with passive collapse of the lung; (b) CT thoracic angiogram revealed evidence of haemothorax, cervical scoliosis but does not reveal any active leak of contrast or a pseudoaneurysm

was sent for analysis such as malignant cells, Acid-fast bacilli (AFB), Adenosine deaminase (ADA), and Genexpert. All reports were within normal limits.

After taking consultation with a cardiothoracic surgeon, intercostal drainage (ICD) was performed and about 1400 ml clotted blood came out. After ICD, the patient became very comfortable, and there was no evidence of re-bleed. Video-assisted semi-rigid thoracoscopy was performed under mild sedation to find out any evidence of abnormal blood vessels, or malignancy, but we could not find any source of the active bleed. In follow-up, the X-ray chest revealed mild evidence of pleural fibrosis on the right side, and the patient has remained comfortable to date.

DISCUSSION

Neurofibromatosis type 1 is an autosomal dominant disease characterized by multiple dermatological disorders and the less frequent manifestations are vascular abnormalities. The expected life of these patients is usually 10 years less than expected; most of the patients have complications between the fifth and sixth decade. There are various causes of death explained in the literature but pulmonary complications are rare. NF1 patients may have lung involvement as lung cancer, interstitial lung disease, and pulmonary hypertension. Complications such as spontaneous hemothorax are rare but potentially lethal. Clinicians should be aware of this syndrome as a possible cause of sudden death in patients with neurofibromatosis. In our patient, we were initially suspecting septicemia or cardiogenic shock, but after seeing the X-ray and clinical examination, we thought this might be a case of malignancy, pyothorax, or a pulmonary embolism. After the imaging studies and diagnostic aspiration, it was confirmed that this is a case of spontaneous hemothorax.

In the existing literature, few cases of spontaneous massive intrathoracic hemorrhage in patients with NF1 have been reported [4]. Miura and colleagues from Japan have reported some cases of spontaneous hemothorax and hemo-mediastinum of various aetiologies in NF1 [5]. Chest wall abnormalities, including vasculopathy of the intercostal artery, have been reported as a cause of spontaneous hemothorax [6]. In this clinical scenario, early recognition is very important for a definitive diagnosis and optimal therapeutic interventions. If during the imaging study, the etiology of bleeding is identified as vascular anomalies like arterial aneurysms, then endovascular embolization is the treatment of choice in a stable hemodynamic situation, as this intervention is less invasive and more effective than surgical intervention [3,7]. In our patient, the findings of NF1 were typical. She has multiple neurofibromas, cervical scoliosis, and café-au-lait spots all over the body.

CONCLUSION

Neurofibromatosis 1 is an autosomal dominant disease that characteristically affects the skin and nervous tissue, and various other organs of the body. In these patients, pulmonary complications are not a well-recognized entity. Spontaneous hemothorax is a very rare, but potentially lethal complication. Any patient of NF1, if presented to the emergency department in a state of shock, hemothorax must be suspected. Early recognition and appropriate investigation strategy are of paramount importance in saving the life of the suffering patient.

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Funding: None; Conflicts of Interest: None Stated.

How to cite this article: Shukla AN, Singh AK, Nigam S. Spontaneous hemothorax: A rare lethal pulmonary complication of neurofibromatosis type 1. Indian J Case Reports. 2021;7(10):433-435.