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CASE REPORT

A MALE PATIENT PRESENTING WITH BULLOUS LUNG DISEASE AND RECURRENT PNEUMOTHORAX

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A 70-years-old male patient never smoker presented to the Chest department, Assiut University hospital, Egypt with acute-onset of dyspnea and left sided chest pain radiating to the back after sudden attack of severe cough.

The patient denies any other Chest complains and had never been exposed to fumes, dusts, asbestos or silica. The patient gives past history of left sided spontaneous pneumothorax 15 years ago which was managed by tube thoracotomy and Tetracycline pleurodesis. His family history was irrelevant.

General examination shows no abnormality except for tachypnea (24 breaths/min) and his blood pressure was elevated. Skin examination shows multiple café-au-lait spots, which were flat, round spots on the skin. In addition, numerous skin nodules characterized by being mobile, soft and not tender following nerves course present at upper trunk (Fig. 1). His local chest examinations show signs of right sided pneumothorax.

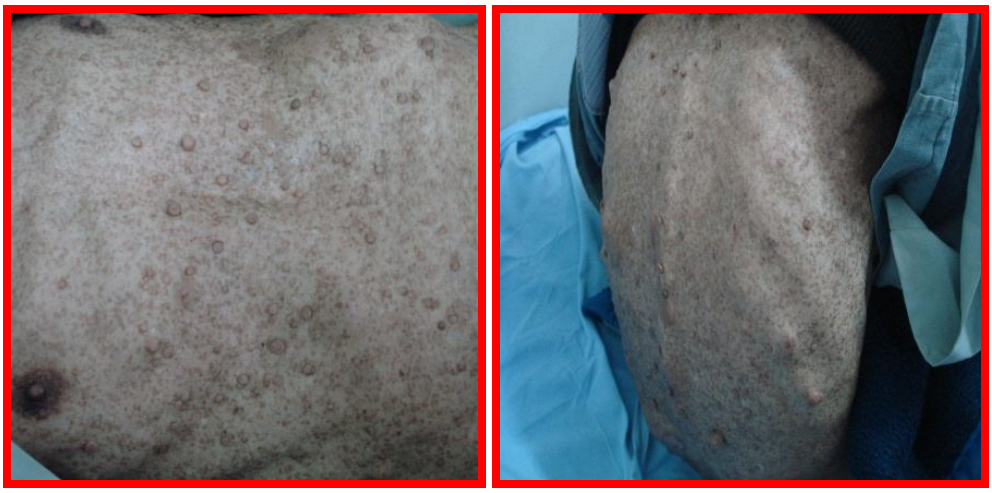

Fig 1. Multiple café-au-lait spots with many skin nodules.

Chest x- ray PA and lateral views revealed right sided pneumothorax (Fig. 2).

The case was initially observed while applying high flow oxygen therapy 60% aiming that the lung expands without any further invasive intervention. A decision to apply tube thoracotomy was to be considered if the initial conservative management failed. Repeated clinical and radiological evaluation was done. After one week, complete lung expansion occurred (Fig. 3). CT scan chest revealed thin walled bullae, predominantly located in the upper parts of the lung. They are different in size, sometimes large and apparently isolated (Fig. 4).

After resolution of pneumothorax a biopsy from one of skin nodules showed neurofibroma.

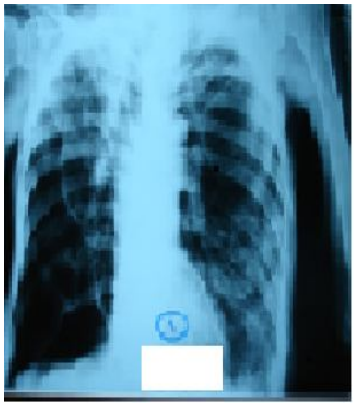

Fig 2. Chest x-ray showing Right sided pneumothorax.

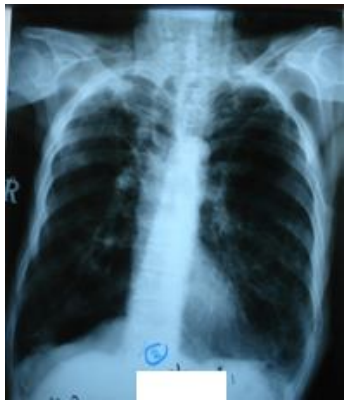

Fig 3. Chest x-ray showing resolution of right sided pneumothorax.

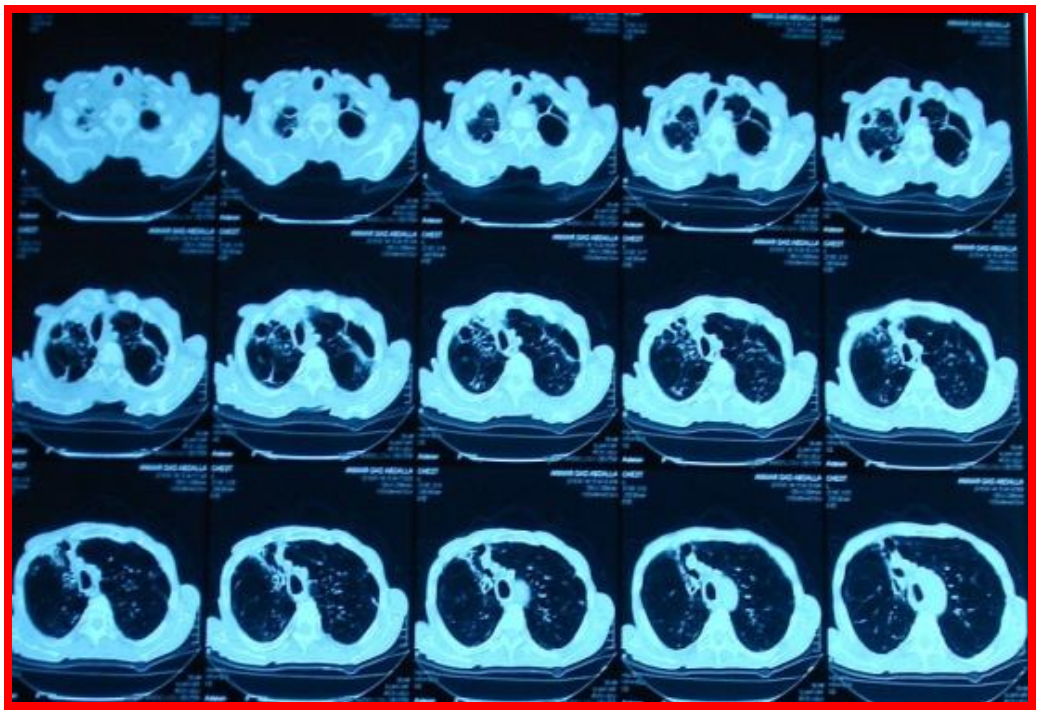

Fig 4. CT scan chest showing multiple apical thin walled bullae.

Final diagnosis: Neurofibromatosis-associated Bullous Lung Disease.

DISCUSSION

Neurofibromatosis (also called von Recklinghausen's disease), an autosomal dominant neurocutaneous syndrome that mainly affects the ectoderm and mesoderm. It is a condition in which noncancerous tumors called neurofibromas grow along nerves in the skin, brain, and other parts of the body. Neurofibromatosis 1 is a subtype of neurofibromatosis. Beginning in early childhood, almost all people with neurofibromatosis 1 have multiple café-au-lait spots, which are flat, round spots on the skin that are the color of coffee with milk. These spots increase in size and number as the individual grows older. Freckles under the arms and in the groin develop later in childhood. Neurofibromatosis 1 is also characterized by the presence of neurofibromas, which are usually located on the skin and often increase in number over time. Neurofibromas may also occur in the spinal cord or along nerves elsewhere in the body. Additional signs and symptoms include high blood pressure and bony changes such as curvature of the spine (scoliosis). Most people with neurofibromatosis 1 have normal intelligence. Fewer than 10 percent are mentally retarded, but about half of affected children have learning disabilities.

In the chest, neurofibromatosis may involve the chest wall, mediastinum and lung parenchyma. In the chest wall, cutaneous neoplasms may appear as nodules on the chest radiograph. Neurogenic neoplasms arising from intercostal nerves away from the spine may give rise to signs of an extrapleural soft tissue mass possibly associated with a notching of the adjacent ribs expressing a pressure remodelling of the ribs. Kyphoscoliosis is common as well as vertebral abnormalities. Middle mediastinal masses are due to thoracic neurofibromas. Diffuse masses often extend down from the thoracic inlet to the level of the hila and may be bilateral. They result from plexiform neurofibromas. These lesions grow slowly and are most commonly asymptomatic. In the lung parenchyma involvement takes the form of fibrosing alveolitis. The chest radiograph shows a reticular pattern or reticulonodular pattern, Kerley lines and honeycombing. Thin walled bullae, predominantly located in the mid upper part of the lung, often asymmetrically, may be seen. They are sometimes large and apparently isolated. Occasionally thoracic neurofibroma may be seen as a solitary pulmonary nodule or an endobronchial tumour.

Differential Diagnosis of Bullous Lung Disease

- Tobacco smoking.
- A1AT deficiency.
- HIV infection.
- IV drug use (ie, methylphenidate, heroin, cocaine, or talc).
- Marijuana smoking.
- Cocaine smoking.
- Autoimmune diseases (ie, hypocomplementemic urticarial vasculitis syndrome, Sjögren disease, Wegener granulomatosis disease, and multisystem autoimmune dysfunction.
- Connective tissue disorders (ie, cutis laxa, Ehlers-Danlos syndrome, and Marfan syndrome.
- Bullous sarcoidosis.
- Idiopathic giant bullous emphysema.
- Birt-Hogg-Dubé syndrome.
- Neurofibromatosis.
- Placental transmogrification of the lung.
- Fabry disease.
- Salla disease.