VOLUME - 11, ISSUE - 10, OCTOBER - 2022 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Radio-Diagnosis

NEUROFIBROMATOSIS ASSOCIATED DIFFUSE CYSTIC LUNG DISEASE

Dr. Divya Jadhav	Resident, Department of Radio-diagnosis Dr.Vithalrao Vikhe Patil, Foundations Medical College Ahmednagar
Dr. Sushil Kachewar	Professor and Head of Department of Radio-diagnosis Dr Vithalrao, Vikhe Patil Foundations Medical College Ahmednagar
Dr. Saurabh Patil	Resident, Department of Radio-diagnosis Dr Vithalrao Vikhe Patil, Foundations Medical college Ahmednagar

ABSTRACT Neurofibromatosis is an autosomal dominant disorder of neuroectodermal and mesenchymal origin, also known as Von Rechlinghausen disorder. It can also occur inherited or sporadically. A mutation of NF1 gene on chromosome 17q11.2 has been recognized as a disease prompter, and the spectrum of clinical phenotype is wide because of gene complexity, size and diversity of exons. In our article we are presenting a case of neurofibromatosis associated with diffuse cystic lung disease.

KEYWORDS : HRCT, Neurofibromatosis, Cystic fibrosis

INTRODUCTION

Neurofibromatosis with diffuse lung disease is characterized by upper lobe cystic and bullous disease and lower lobe fibrosis[1]

Neurofibromatosis Type 1(NF) or Von Recklinghausen's disease is an autosomal dominant neurocutaneous disorder with different clinical expression comprising of café-au-lait spots, neurofibromas, lisch nodules, axillary freckling, optic nerve glioma, kyphoscoliosis.[2]

Thoracic manifestations mainly include mediastinal masses such as neurofibroma, lateral thoracic meningocele, extra-adrenal pheochromocytoma and lung parenchymal disease such as diffuse interstitial fibrosis in lower zone, upper lobe bullae formation and secondary pulmonary arterial hypertension. [3]

NF-1 gene locus is located on chromosome 17q11.2 NF has a prevalence of one in 3,000 and in 30–50% of cases there is no family history of the disease. These sporadic cases probably arise from (usually paternal) germ cell mutations. [4]

Case Presentation

A 70 year old male, presented with difficulty in breathing, cough and fever for last 15 days. Patient had no history of smoking. No pulmonary function test was performed. Patient's vitals were normal and oriented to time, place, person. His physical examination revealed soft multiple cutaneous neurofibromas over entire body. On systemic examination revealed reduced air entry bilaterally and inspiratory crackles.

Chest radiograph show large basal left lower lobe cyst and computed tomography scan of thorax was recommended. Mild ground glass opacities with multiple scattered thin walled cysts in both lung field ranging from 5mm – 45 mm, most prominent in right upper and in left lower lobes. Thus above findings raised suspicion of Neurofibromatosis associated cystic lung disease.

DISCUSSION

Pulmonary manifestations of NF-1 show characteristic upper lobe involvement usually presenting in fifth - sixth decade with predilection to history of smoking. [5]

Pathology- Neurofibromin in a protein encoded by tumor suppressor gene NF-1 that downregulates Ras activity. [6] Thus mutated gene causes inadequate neurofibromin production thereby proliferative cell growth with increased collagen deposition however exact pathophysiology is still unknown[9] In lung tissue there is increased lymphocytic infiltration around alveolar septa causing bronchiolar dilatation leading to cyst formation[7].

Presentation-

Patient present with non-specific symptoms such as cough, chest pain, dyspnea with an incidental finding on HRCT. Other abnormalities in HRCT seen are bullae formation, reticular abnormalities, ground glass opacities, cysts and emphysema. Although there no profound evidence of smoking leading to cystic lung disease in NF yet it manifest and increase the severity of disease progression. Complications- respiratory failure, spontaneous pneumothorax

CONCLUSION

Neurofibromatosis is an autosomal dominant disorder of neuroectodermal and mesenchymal origin. [8]It is frequently associated with lung involvement. In our article we have presented a rare case of neurofibromatosis which is associated with lung involvement showing diffuse cystic lung disease. However many studies show relevance of smoking and cyst formation in such cases, our patient had no history of smoking thus suggesting that lung involvement is not always secondary to it.

X-ray is helpful as initial diagnostic modality to diagnose the pathology. HRCT helps in confirmation of X-ray findings and detailed evaluation of the localization and characteristic of disease findings with consistent follow up.

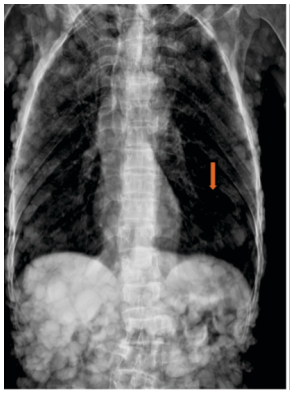


Fig. 1 Chest X-ray AP view showing lower lobe cyst.

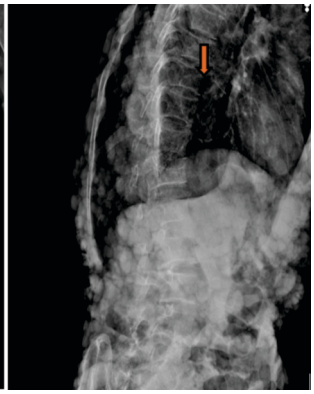


Fig 2 chest X-Ray lateral showing cyst

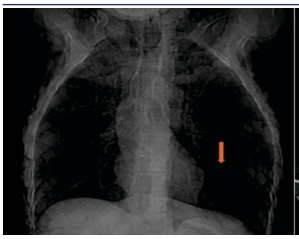


Fig no 3 HRCT scout showing cystic bullae

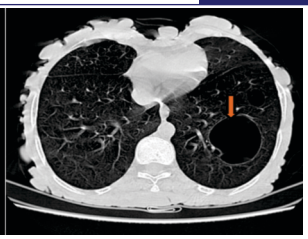


Fig no 4 HRCT axial large rounded lung cyst involving left lower lobe.

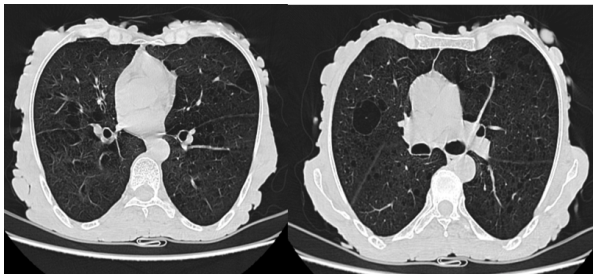
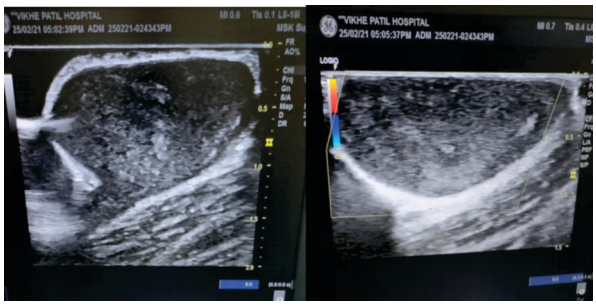
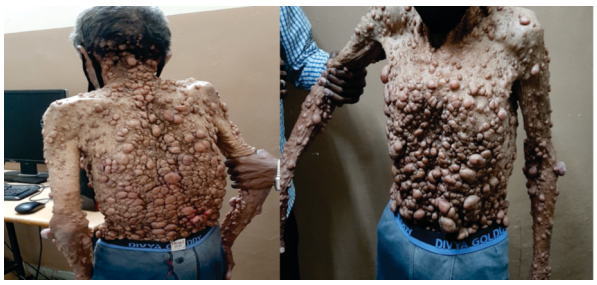


Fig no 5,HRCT Image showing scattered small well-defined round cysts in predominantly upper lobe.



Ultrasound image showing well defined cysts with echogenic content within suggestive of neurofibroma's over superficial layer.



Pictorial depiction of neurofibromas over patient's body.

REFERENCES -

1. Júnior SF, Zanetti G, de Melo AS, Souza Jr AS, Souza LS, Meirelles GD, Irion KL, Hochhegger B, Marchiori E. Neurofibromatosis type 1: State-of-the-art review with emphasis on pulmonary involvement. Respiratory Medicine. 2019 Mar 1;149:9-15
2. Pinson S, Wolkenstein P. La neurofibromatose 1 (NF1) ou maladie de Von Recklinghausen. La revue de médecine interne. 2005 Mar 1;26(3):196-215.
3. Zamora AC, Collard HR, Wolters PJ, Webb WR, King TE. Neurofibromatosis-associated lung disease: a case series and literature review. European Respiratory Journal. 2007 Jan 1;29(1):210-4.
4. Riccardi VM. Von Recklinghausen neurofibromatosis. *N Engl J Med* 1981;**305**:1617–1627.
5. Louza GF, Zanetti G, Marchiori E. Neurofibromatosis type I with pulmonary involvement. Arch. bronconeumol.(Ed. impr.). 2018:106-7.
6. Cichowski K, Jacks T. NF1 tumor suppressor gene function: narrowing the GAP. Cell. 2001 Feb 23;104(4):593-604.
7. Pulmonary hypertension associated with neurofibromatosis type 1. Marie J, Girerd B, Jaïs X, et al. Eur Respir Rev. 2018;27:149.
8. Ozcinar B, Aksakal N, Agcaoglu O, Tukenmez M, Ozemir IA, Barbaros U, Colak N, Erbil Y. Multiple gastrointestinal stromal tumors and pheochromocytoma in a patient with von Recklinghausen's disease. International Journal of Surgery Case Reports. 2013 Jan 1;4(2):216-8.
9. Dehal N, Gastelum AA, Millner PG. Neurofibromatosis-Associated diffuse lung disease: a case report and review of the literature. Cureus. 2020 Jun 29;12(6).