ISSN- O: 2458 - 868X, ISSN–P: 2458 – 8687
Index Copernicus Value: 49. 23
PubMed - National Library of Medicine - ID: 101731606

International Journal of Medical Science and Innovative Research (IJMSIR)
IJMSIR : A Medical Publication Hub
Available Online at: www.ijmsir.com
Volume – 4, Issue – 1, January - 2019, Page No. : 115 - 117

Case Report: Familial Pneumothorax

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Type of Publication: Case Report

Conflicts of Interest: Nil

Introduction

Familial pneumothorax is a rare disorder. Approximately 10% of patients of spontaneous pneumothorax have positive family history. Familial pneumothorax was first reported by Farber3 in 1921. It can happen in alpha1 antitrypsin deficiency, marfans syndrome, Tuberous sclerosis, cystic fibrosis and Birt- Hogg- Dube (BHD) syndrome. We report three generations of a kashmiri family who suffered spontaneous pneumothoraces in the absence of other features of the BHD syndrome, and were found to have lung cysts. Genetic analysis in this family could not be done due to financial constrains.

Case History

A 28-yr-old female non smoker presented with sudden onset of dyspnea and chest pain from 4 days. Clinical examination revealed left sided decreased chest movements , hyper-resonance in suprascapular, infrascapular, and inframammary areas,and absent breath sounds. Systemic examination was normal except tachypnoea with normal hemodyanamics and saturation of 95% . Chest radiograph showed a left-sidedpneumothorax. An intercostal drain was inserted , High resolution CT scan of chest further confirmed pneumothorax . However, a detailed history suggested that many other members of patient's family had also suffered spontaneous pneumothorax.Her maternal cousins both females and males were affected and diagnosed with a pneumothorax. The index patient's uncle presented with a left-sided pneumothorax at age 45 yrs. A chest tube was placed and pneumothorax resolved. Her four maternal cousins were affected between ages of 20 to 30 yrs four females from same parenthood were affected , her two cousins from maternal side with both being males were also affected.

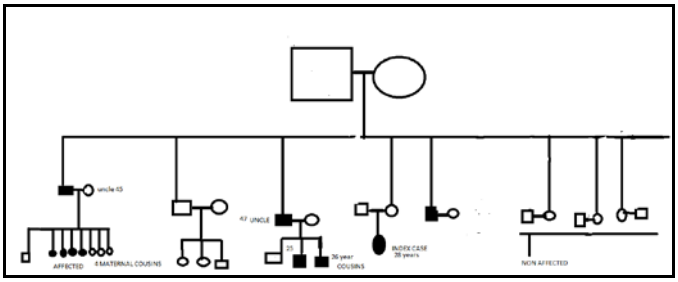

Fig 1.0 shows the family pedigree of the index case.

Radiological screening by high-resolution computed tomography (CT) scan of the chest was done in the patient (fig 1.2) and Multiple pulmonary cysts were found in upper lobe of both lungs in the index case. Cysts were distributed throughout all lobes. The cysts were mostly thin walled, with a few cysts having a thickness of up to 2 mm, or no discernable wall. The largest cyst in our series was 5-7 mm in diameter. The lung parenchyma in between these cysts was normal. In the family there was no marfanoid habitus or any evidence of tuberous sclerosis. Additional investigations were offered to our patients to detect related genetic mutations however patients could not afford.

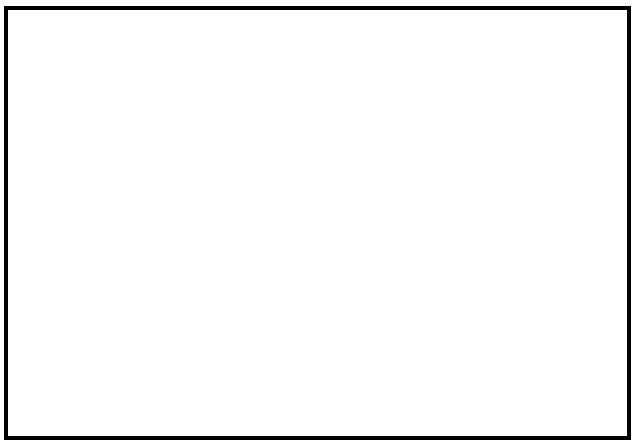

fig 1.1: chest radiograph of index case showing left sided pneumothorax

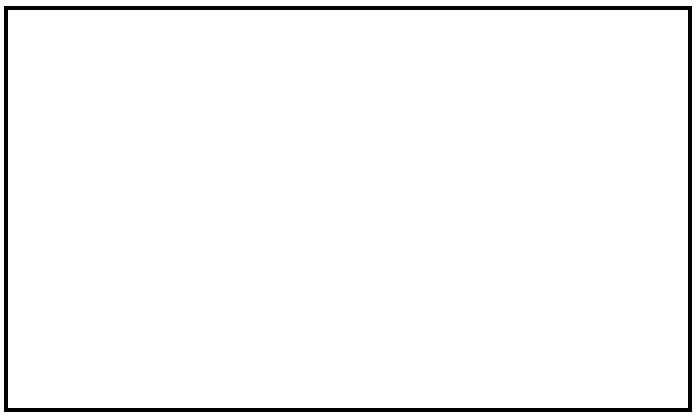

Fig 1.2: High resolution computed tomography scan of chest showing moderate hydro- pneumothorax on left side of chest,few apical subpleural cysts are found

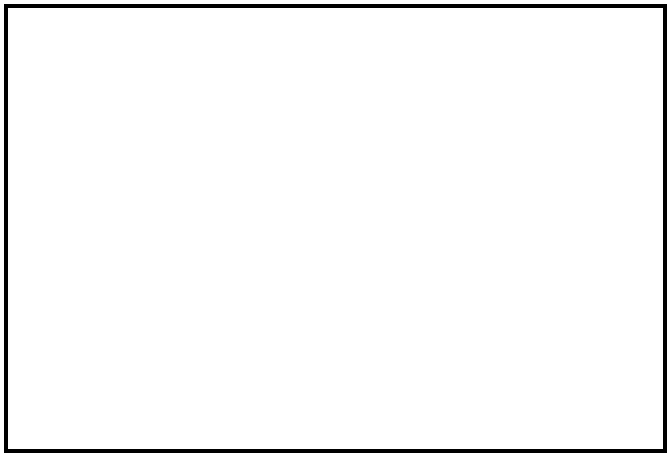

Fig 1.3: Radiograph chest of female maternal cousin of patient showing pneumothorax left upper zone

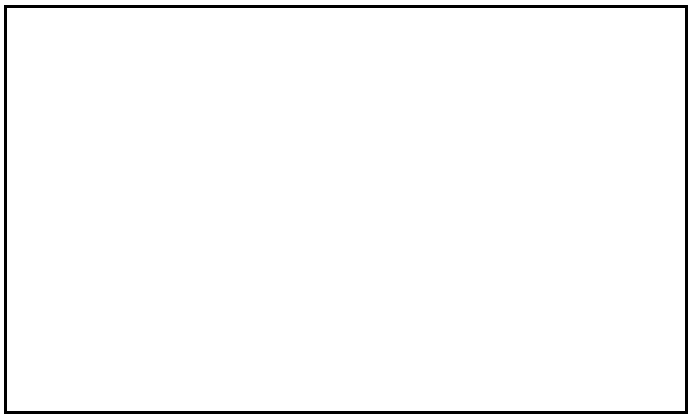

Fig. 1.4 CT chest of 22 year male cousin of pateint

Discussion

Spontaneous pneumothorax often affects tall individuals. 10% of these spontaneous pneumothorax patients have positive history. Birt,Hugh, and Dube in 1977 described a rare autosomal dominant condition of familial pneumothorax associated with renal, colonic neoplasms and benign tumors of hair follicles due to mutations of folliculin (FLCN) gene.[4] It is important to elicit family in all patients of spontaneous pneumothorax.[1,2,3] Positive family history may indicate this rare syndrome, where in there is seven fold increased risk of developing renal neoplasms and 50 fold increased risk of pneumothorax. Prophylactic pleurodesis may be warranted in some affected families. Familial pneumothorax is not a single entity but many genetic disorders like Birt-Hugg-Dube[5,6]

syndrome, Tuberous sclerosis, Lymphangiomyomatosis (LAM), Marfans syndrome, Loeys- Dietz syndrome (TGF beta receptor defect), Ehler Danlos syndrome, alpha 1 antitrypsin deficiency, Homocysteinemia may present as familial pneumothorax. These can have severe extrapulmonary manifestations ranging from renal cancer to aortic rupture. Since pneumothorax is a common early manifestation of these life threatening diseases, it offers an important opportunity to make diagnosis that can facilitate precision medicine. A high level of clinical suspicion is necessary for making early diagnosis.

Conclusion

In conclusion, identifying families with spontaneous pneumothorax is clinically important, as these individuals and their relatives are at increased risk of developing malignancies. Identifying and reporting the genotype will contribute towards understanding the genotype–phenotype associations and in the future, may help explain the molecular basis of cystic lung diseases.

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