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A CASE OF CYSTIC LUNG DISEASE ASSOCIATED WITH MARFANS SYNDROME- A CASE REPORT

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Abstract : Various pulmonary problems have been described in Marfan's syndrome. Here we are having 29 years old male patient who presented with 4 years of progressive dyspnea and cough with history of recurrent respiratory infections since childhood, patient had marfanoid features and features of cor pulmonale, investigations revealed diffuse cystic lesions in both lung fields. Cystic lung disease in marfans syndrome is very rare. We report a case of cystic lung disease in a marfans syndrome due to its rarity of presentation.

Keyword : Marfans syndrome, Cystic lung disease, Ghents criteria.

INTRODUCTION

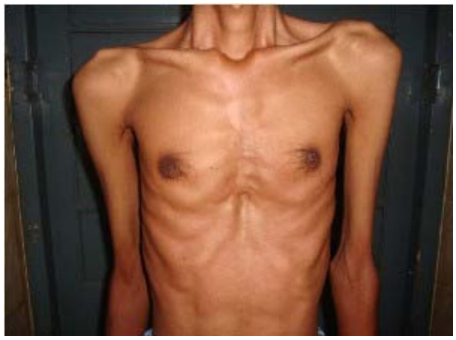
Marfan syndrome is a genetic disorder of the connective tissue transmitted as an autosomal dominant trait usually presents with skeletal, cardiovascular, and ocular abnormalities. The basic pathological abnormality in this disorder affects the supportive elements of the connective tissue. Various pulmonary abnormalities have been described like apical bullous emphysema, spontaneous pneumothorax, cystic lung disease. This is a case report of 29 yrs old male patient, cystic lung disease with marfans syndrome.

CASE REPORT

This is a case description of a 29 yrs old male unmarried patient who is a Goldsmith. Presented to our hospital with complaints of Cough and Breathlessness for the past 4 yrs. Cough was insidious in onset, progressive in nature, associated with minimal mucoid sputum, no h/o hemoptysis. Breathlessness was insidious and progressive in nature, progressed from MRC grade 1 to grade 3. No history of orthopnea, paroxysmal nocturnal dyspnea, no history of palpitations chest pain, facial puffiness, right upper abdominal pain, abdominal distension, lower limb swelling, Fever, Jaundice, Diarrhea. History of Loss of appetite and loss of weight present. History of recurrent respiratory tract infection present since childhood.4 years back, patient presented for similar complaints and was diagnosed as sputum negative pulmonary

tuberculosis and was started on ATT patient took antituberculous treatment for 6 months and lost follow up. Personal history- patient is non smoker and not an alcoholic.

Family history- born of non consanguineous marriage, both parents are not alive. Father was also tall and thin like him who died at the age of 54 yrs probably due to stroke. Mother also died at the age of 54yrs due to some kidney disease. 3 children, other 2 siblings are normal.



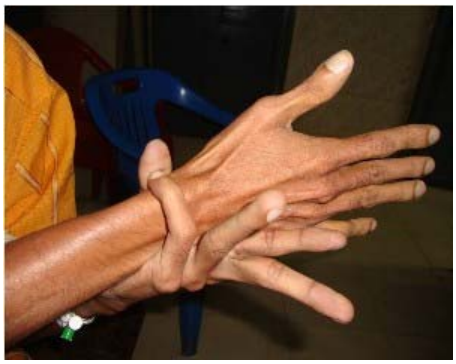


Fig-1

Fig-2 Fig-3

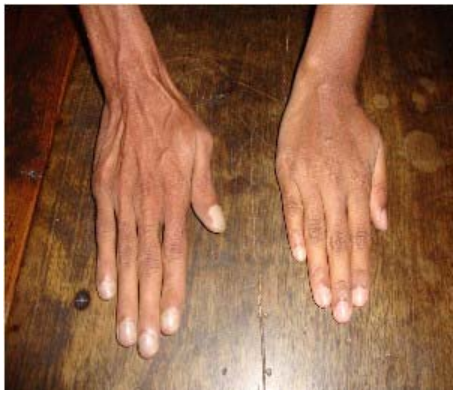


Fig-4

Fig-5

On Examination patient was Conscious, oriented, afebrile, poorly built and nourished cyanosis present, grade 2 pan digital clubbing was present, no pedal edema, no generalized lymphdenopathy.Pulse-110/min, Blood pressure -90/70mm/hg, respiratory rate 27cycles/min, jugular venous pressure was normal. The following Skeletal deformities were present pectus excavatum+ (Fig-1),wrist sign+ (Fig-2),thumb sign+(Fig3) Arachnodactyly+ (Fig4),hypermobilityofmetacarpophalangeal joints +,high arched palate+ Armspan180cms>height 17ocms the ratio was 1.05(Fig-5),Upper segment 79cms<LS 91cms the ratio was 0.86.No striae, no hernias, No kyphosis, scoliosis, pes planus. Respiratory system-Patient Tacypneioc, accessory

muscles of respiration is acting. Trachea is central in position, reduced movements in the right upper hemithorax ,apical impulse is seen in the left 5th intercoastal space in midclavicular line, right side drooping of shoulder was present scapula was prominent on right side, pectus excavatum present, no kyphosis or scoliosis. Auscultation revealed bilateral crepitations and wheeze in all over the lung fields. Cardiovascular system revealed loud second heart sound in pulmonary area, no murmur. Abdominal system and nervous system examination revealed no significant abnormality. Ophthalmic examination revealed myopia in both eyes 6/12, hyperemic fundus. slit lamp examination revealed deep anterior chamber, posterior subcapsular cataract, lens in situ, no dislocation of lens.

His blood investigations revealed, hemoglobin -18.6 gm%, hematocrit -58.9%,Total WBC count 67OO,DifferentialLeucocyte Count P56,L40,E2,Platelets-1.8lakh/cu mm, Random blood sugar-124mg%,Urea-48mg%,Creatinine -1.2mg%,Urine routine – normal, Arterial blood gas analysis showed-respiratory alkalosis. **HIV ELISA**- Non reactive, Ultrasound abdomen**-** normal, **ECG** showed sinus tachycardia, P pulmonale ,Right axis deviation and right ventricular hypertrophy, Sputum for AFB**-**negative, Alpha one antitrypsin levels (Nephelometry method) 186 (Normol 84 -163).

ECHOCARDIOGRAM- Right atrium right ventricle minimally dilated, Pulmonary hypertension mild, Right ventricular systolic pressure 40 mm hg, Tricuspid regurgitation mild, normal Left ventricular function, Aortic root and sinuses normal. **CHEST X RAY** (Fig-6)- diffuse bilateral miliary mottling, with minimal fibrosis in the right upper zone.

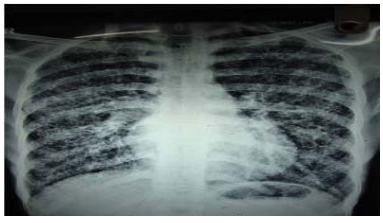


Fig-6

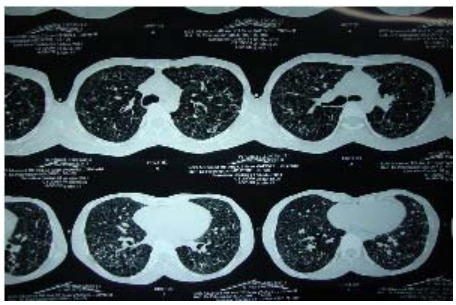
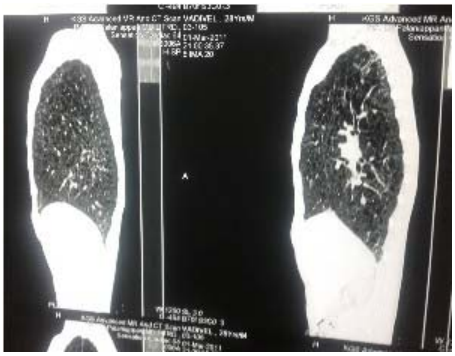


Fig-7



CT THORAX (Fig 7,8) - bilateral diffuse uniformly distributed cysts in both lung fields no hilar or mediastinal lymphadenopathy,pleura normal. **X RAY HAND** showed **METACARPAL INDEX**-9.76/0.84=11.61(>8.4 Indicates-Arachnodactyly).

He was treated with oxygen, diuretics, antibiotics and other antifailure measures for cor pulmonale. Patient was discharged and adviced to get home oxygen therapy. Patient after 1 week of discharge, succumbed to his illness probably due to severe respiratory failure. **DISCUSSION** Marfan syndrome is a genetic disorder of the connective tissue transmitted as an autosomal dominant trait**,** but about one-fourth of patients have sporadic new mutations. It is noteworthy for its worldwide distribution, relatively high prevalence about 1 in 3000/5000 births in most racial

and ethnic groups, clinical variability, and pleiotropic manifestations, some of which are life threatening[1]. About three quarters of patients have an affected parent; new mutations account for the remainder. Marfan syndrome is fully penetrant with marked interfamilial and intrafamilial variability. No sex predilection is known[1]. **PATHOPHISIOLOGY** Marfan syndrome results from mutations in the fibrillin-1 (*FBN1) gene on chromosome 15, which encodes for the glycoprotein fibrillin. Fibrillin is a major building block of microfibrils, which constitute the structural components of the suspensory ligament of the lens and serve as substrates for elastin in the aorta and other connective tissues. Progressive aortic dilatation and eventual aortic dissection occur because of tension caused by left ventricular ejection impulses. Likewise, deficient fibrillin deposition leads to reduced structural integrity of the lens zonules, ligaments, lung airways, and spinal dura. Whether the cystic changes are congenital in origin or whether they arise from premature degeneration of pulmonary parenchyma is not clear. The precise cause of the gross pulmonary disease remains uncertain; it may be a consequence of abnormal collagen, causing flaccidity of the walls of the terminal bronchioles during expiration and thus obstruction and air trapping. It is important to recognise this complication of Marfan's syndrome as it may lead to spontaneous*pneumothorax.

CLASSIFICATION

MFS was initially characterized by a triad of features: (1) skeletal changes that include long, thin extremities, frequently associated with loose joints (2) reduced vision as the result of dislocations of the lenses (ectopia lentis) and (3) aortic aneurysms[1]. Analyses of patients without mutations in the *FBN1 gene identifies mutations in the gene that encodes transforming growth factor receptor 2 (TGFBR2). These patients were classified as type II MFS. Patients with type II MFS were similar to patients with type I MFS but lacked the ocular changes. In subsequent studies, mutations in both TGFB2 and the closely related TGFB1 gene were found in patients with a new syndrome referred to as Loeys-Dietz aneurysm syndrome (LDAS). The patients with LDAS presented with aneurysms of the ascending aorta, tortuous arteries, cleft palate, and hypertelorism , and the presence of contractures with some of the signs of Osteogenesis*Imperfecta suggests Congenital Contractual Arachnodactyly[1]. · An international panel has developed a series of "Ghent criteria" that are useful in classifying patients.

DIAGNOSTIC (GHENTS) CRITERIA FOR MARFANS SYNDROME[2]

If family history is not contributory, index case diagnosis requires major criteria in 2 organ systems and involvement in third. Alternatively, if a mutation known to cause marfans syndrome is present, Index case requires 1 major criterion in 1 organ system and involvement in second. Diagnosis of index case's relative requires 1 major criterionin family history.1 major criterion in 1 organ system, and involvement in a second **.** Minor criteria are used only to score organ system involvement and do not count towards the diagnosis.

Our patient has got following major criteria in **MUSCULOSKELETAL SYSTEM**-Thumb sign, wrist sign, pectus excavatum, Arm span 180 cms>height 170 cms the ratio was1.05, Upper segment 79cms<LS 91cms the ratio was 0.86, following minor criteria Arachnodactyly, hypermobility of metacarpophalangyl joint, high arched

palate. In **EYES** our patient has got following minor criteria Myopia, Deep anterior chamber. In **LUNGS**- Cystic lung disease with Positive family history.

Pulmonary manifestations in Marfan's syndrome;

Interstitial parenchymal disease and honeycombing. Diffuse and apical bullous emphysema. Congenital malformations of the bronchus, bronchiectasis. Spontaneous recurrent pneumothorax. Lung cysts and bullae are uncommon, however. In a recent case reports A case of diffuse cystic changes on the chest radiograph which were mistaken and treated as pulmonary tuberculosis have been described.(13) MANAGEMENT

Multifaceted.Routine and serial clinical examination and ECHO. Prophylaxis before any procedure.Long acting beta blocker propronolol and atenolol-reduce pulse rate, systolic blood pressure and prevent aortic complications and arrhythmias. ACEI AND ARB to prevent aortic complications.[7] Surgical correction of aortic and mitral valves.[1]

ABBREVATIONS

ECHO-Echocardiogram MFS-Marfan syndrome FBN-Fibrillin ECG-Electrocardiogram ACEI- Angiotensin converting enzyme inhibitors ARB-Angiotensin receptor blockers N/A-Not applicable MRI-Magnetic resonance imaging AFB-Acid fast bacilli CT-Computed tomography.

CONCLUSION;

This is a case of cystic lung disease associated with marfans syndrome, we are presenting this case due to its rarity and poor prognosis in young age and absence of proper treatment.

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