INTRODUCTION
Marfan syndrome is an autosomal dominant, multisystem connective tissue disease, associated with a mutation in fibrillin, and occasionally a mutation in TGFBR1 or 2.3,4 The cardinal features of this condition revolve around cardiovascular, ocular and skeletal system.5 Prevalence is 1 per 5000 population and 26% cases have no family history.6, 7 Main features include aortic root dilatation, chest wall deformities, long slender fingers, lens dislocation and myopia. Diagnosis is usually made by using the revised Ghent criteria which includes a detailed clinical examination.8 Even though the life expectancy has increased from 40 to 70 years2, with early diagnosis of complications especially cardiovascular; they continue to suffer significant morbidity. Pulmonary manifestations which the literature describes include restrictive lung diseases, sudden lung collapse (spontaneous pneumothorax), emphysema, asthma and sleep apnea.10 Association of Marfan syndrome with empyema and tuberculosis is not described in the literature.

CASE REPORT
A 30 year old female, presented to the emergency department with acute onset of dyspnoea for 2 days; fever, high grade, with chills and rigors, intermittent lasting for a few hours a day and partly relieved by medications from local doctors for 2 days; cough which was dry initially, but later productive of whitish sputum for 2 days and chest pain which was dull aching, vague, more on right side of chest for 2 days. She had no history of orthopnea, paroxysmal nocturnal dyspnoea or palpitations.

Her past history was significant for sputum positive pulmonary tuberculosis a year and a half ago, for which she was put on Anti-Koch’s therapy (Category I AKT under Revised National Tuberculosis Control Programme, RNTCP) and she was sputum negative within the first two months of treatment. She continued and finished her full course of Category I AKT for 6
months. 4 months ago she had similar symptoms of dyspnoea, fever and cough, for which she went to a local tertiary care hospital, where right sided fluid was diagnosed on Chest X-Ray and chest ultrasound, but ultrasound guided tapping did not aspirate any fluid. She withheld any further treatment thereafter and went home on oral antibiotics, from which she improved symptomatically, but there was no follow up. Her family history was negative for any major ailments as well as any genetic disorder. Clinically, her blood pressure was 104/70, with a heart rate of 110, and tachypnea at 28/min. Her O₂ saturation was 86% on room, air and 95% with nasal oxygen. On examination, she had thin, slender fingers, pectus excavastum and a visually tall stature. Her height was 167 cms (5’ 4”) and arm span of 179 cms (5’ 9”). Upper to lower segment ratio of 0.81 and arm span to height ration of 1.07. Her chest wall showed pectus excavatum. Her air entry was absent on right basal and lower zone region, both anteriorly and posteriorly. Her other system examination were all normal. Her laboratory findings are as follows:

Her Complete Blood Count (CBC) showed hemoglobin of 11.9 gm/dl; with total count raised to 13,300 /cu. mm with differential of N 94%, L 04%. Her X-Ray showed slight haziness in right basal lung field. ECG was suggestive of P-pulmonale with T inversion in V3 to V6 with persistence of S wave till V6. Her ultrasound chest showed mild free fluid with thick internal separations with the underlying lung consolidated. Pulmonary function tests were normal with peak expiratory flow rate (PEFR) 120 /min. HRCT Chest suggestive of:

- Loculated right mild pleural effusion with enhancing pleural thickening representing empyema.
- Collapse of right middle lobe was seen.
- Thin walled cavities in bilateral upper lobes and apical segment of right lower lobe.
- Dilated main pulmonary artery may represent pulmonary arterial hypertension.
- Calcification in apical segment of right lower lobe representing granuloma.

Her sputum stain was suggestive of gram positive coccus in pairs (p/o Streptococcus pneumonia and thick gram negative bacilli p/o capsulated organisms; cultures were however negative) and her sputum acid fast bacilli (AFB) was negative. 2D-Echo report was suggestive of bellowing of AML into LA in systole, her Z-score for aortic root being 2.8.

X-Rays of wrists were done with metacarpal index of 8.8. X-Ray Pelvis with bilateral hips showed angle of Wiberg of 42. Her thumb’s sign (Walker) and Wrist sign (Steinberg Sign) are positive. Her chest wall is suggestive of pectus excavatum.

According to revised Ghent Criteria, Systemic calculated from above features was 8 and an aortic root Z score of 2.8, a diagnosis of Marfan Syndrome was made, with her HRCT Chest showing changes of empyma and cavity with granulomas suggestive of tuberculosis.

She was put on injectable higher antibiotics, started on Category II Anti-Koch’s’ treatment and discharged on oral antibiotics with close follow up. Her only child a son was screened for tuberculosis as well as features of Marfan syndrome, both of which were negative.
DISCUSSION

Marfan syndrome is associated with an array of systemic involvement. Ocular manifestations are subtle, skeletal manifestations are obvious on observations and cardiac manifestations are life threatening. Ghent’s (old) criteria did not have any major pulmonary criteria, only minor, when it came to systemic involvement and scoring.11 This shows that pulmonary manifestations are underscored by other system involvement. Recurrent and severe respiratory infections are something which has not been in limelight when it comes to pulmonary complications. A patient with Marfan syndrome has all possible reasons to develop recurrent infections, these include: a poor skeletal structure of spine (abnormal spine curvature; scoliosis > 20°, spondylolisthesis, pectus excavatum or carinatum), with pulmonary blebs and emphysematous chest and occasionally development of bronchiastasis. With all these reasons, the lungs become a perfect candidate for the organisms to flourish. So, a lot of attention has to be paid when a Marfan syndrome patient presents with any respiratory signs and symptoms. Any upper respiratory tract infections should be aggressively treated and special care and follow up is required for such patients. This also means that before any procedures or surgeries, adequate respiratory care should be taken including antibiotics and physiotherapy.

REFERENCES: