Case report

Spontaneous Pneumothorax in a Patient with Marfanoid Habitus

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Abstract

A 39-year-old smoker with Marfanoid habitus presented with sudden onset shortness of breath. The diagnosis of right-sided primary spontaneous pneumothorax was made. It was successfully managed by drainage with inter costal tube. Presence of Marfanoid phenotype and smoking will increase the risk of spontaneous pneumothorax.

Key words: Spontaneous pneumothorax; Marfanoid habitus;Chronic smoking

Funding: None

Competing interest: None

Received: 21 January2016 **Accepted revised version:** 03 July 2016 **Published:** 01 September 2016

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Cite this article as: Basnayake BMDB, Kannangara T, Welagedara RL. Spontaneous pneumothorax in a patient with marfanoid habitus. *Anuradhapura Medical Journal* 2016; **10**: 17-20.
DOI: http://dx.doi.org/10.4038/amj.v10i1.7600

Introduction

Pneumothorax occurring without any trauma or obvious precipitating factor is called spontaneous pneumothorax (SP) (1). There are two types of SP; primary spontaneous pneumothorax (PSP) occurs in patients without any clinically apparent underlying lung disease and secondary spontaneous pneumothorax as a complication of pre-existing lung pathology (2). PSP is commonly seen in chronic smokers and patients with Marfanoid habitus (1, 3). We report a case of right-sided primary spontaneous pneumothorax in a chronic smoker with Marfanoid habitus.

Case presentation

A 39-year-old previously healthy man presented with sudden onset shortness of breath for two days duration. There was no history of chest pain, cough or trauma to the chest. He is a chronic smoker (20 cigarette pack years). On examination he had Marfanoid features; height was 178.2 cm and arm span was 194 cm and arm span to height ratio was more than 1.05 (Figure 1). The lower body segment measured 101 cm, with an upper-lower body ratio of 0.76. Wrist (Walker) and thumb (Steinberg) signs were positive. He had a long thin face, high arched palate, pectusexcavatum and arachnodactyly.

Figure 1 **Marfanoid habitus: tall stature and dolichostenomilia**

Joint hypermobility or hyperelasticity of the skin were not evident. On further questioning, his father and other siblings had similar body habitus. Respiratory rate was 22-cycles per minute & trachea was in the midline. There was reduced air entry and hyper-resonance over the right hemithorax. Oxygen saturation was 98% on air. Right-sided pneumothorax was confirmed by chest radiography (Figure 2). Inter costal tube with underwater seal was inserted with subsequent resolution of pneumothorax. There were no cardiac murmurs. Ophthalmologic examination revealed no evidence of lens dislocation or cataract. Abnormalities such as aortic root dilatation, mitral valve prolapse or mitral regurgitation were not present in 2D echocardiogram. All the other investigations were normal. Mantoux test was negative. After two days of hospital stay he was discharged. No further recurrence of pneumothorax was observed during the follow up clinic visit.

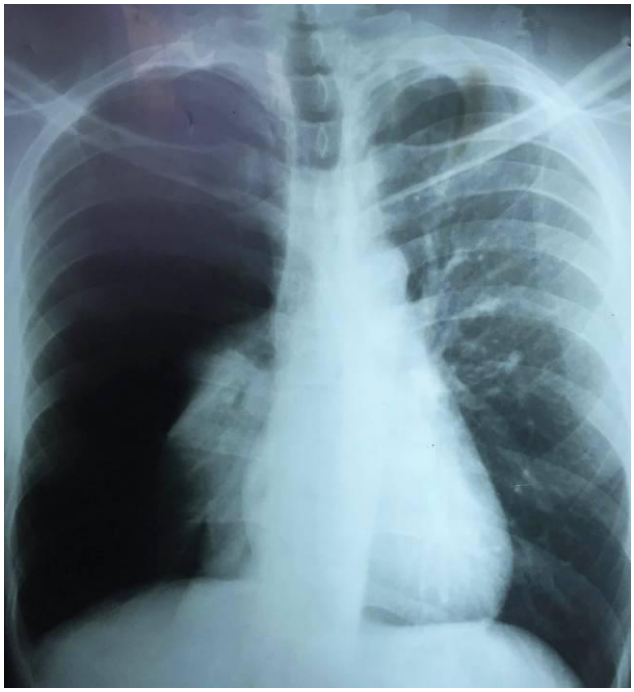

Figure 2 **Chest radiograph showing right sided pneumothorax**

Discussion

The incidence of PSP is 7.4/100 000 for men and 1.2/100 000 for women (3). Lifetime risk of developing pneumothorax in smokers is 12% compared to only 0.1% in non-smokers (4). Marfanoid habitus is associated with Marfan syndrome, Shprintzen-Goldberg syndrome, Lujan-fryns syndrome and congenital contractual arachnodactyly (5). The revised Ghent criteria (Box 1) are used currently to diagnose Marfan syndrome (6). Our patient had a total systemic score of 7 (Wrist AND thumb sign–3, pectusexcavatum–1, Pneumothorax–2, Reduced upper-lower body ratio AND increased arm span to height ratio AND no severe scoliosis –1) indicating systemic involvement of Marfan syndrome. Recognized pulmonary manifestations of Marfan syndrome are spontaneous pneumothorax (4–11%), pectusexcavatum, scoliosis, emphysema, bullae and apical blebs. The presence of apical blebs, bullae, abnormal connective tissue constituents in the lung parenchyma, or increased mechanical stresses in the lung apices due to the tall body habitus will contribute to and increased risk of developing spontaneous pneumothorax (7,8). Our patient had two major risks; Marfanoid habitus and chronic smoking leading to the development of PSP. Treatment options for PSP are observation for small closed pneumothoraxes, simple aspiration as first line treatment for all primary pneumothoraxes requiring intervention, and intercostal tube drainage if simple aspiration and drainage is unsuccessful. Due to the increased risk of recurrence, they

should be advised on cessation of smoking and avoidance of scuba diving, fast ascents in elevators and playing brass instruments (8, 9).

Conclusion

In a male smoker with Marfanoid phenotype sudden onset shortness of breath should alert the presence of pneumothorax.

Consent

Informed written consent was obtained from the patient for publication of this case report and images. A copy of the written consent is available for perusal with the Editor-in-Chief of this journal.

Acknowledgments

We thank Dr. PNS Premathilake for reading through the manuscript and the invaluable corrections she made to it.

Box 1 **Revised Ghent criteria for the diagnosis of Marfan syndrome**

In the absence of a family history:

(1) Ao (Z ≥ 2) AND EL = MFS
(2) Ao (Z ≥ 2) AND FBN1 = MFS
(3) Ao (Z ≥ 2) AND Syst (≥7 points) = MFS
(4) EL AND FBN1 with known Ao = MFS

In the presence of a family history:

(5) EL AND FH of MFS (as defined above) = MFS
(6) Syst (≥ 7 points) AND FH of MFS (as defined above) = MFS
(7) Ao (Z≥2 above 20 years old, ≥ 3 below 20 years) + FH of MFS (as defined above) = MFS

Systemic score

- Wrist AND thumb sign –3 (wrist OR thumb sign –1)
- Pectuscarinatum deformity –2 (pectus excavatum or chest asymmetry –1)
- Hindfoot deformity –2 (plain pes planus –1)
- Pneumothorax –2
- Duralectasia –2
- Protrusioacetabuli –2
- Reduced US/LS AND increased arm/height AND no severe scoliosis –1
- Scoliosis or thoracolumbar kyphosis –1
- Reduced elbow extension –1
- Facial features (3/5) –1 (dolichocephaly, enophtalmos, down slanting palpebral fissures, malar hyoplasia, retrognathia)
- Skin striae –1
- Myopia > 3 diopters –1
- Mitral valve prolapse (all types) –1

Maximum total: 20 points; score ≥ 7 indicates systemic involvement

Ao = aortic diameter at the sinuses of Valsalva above indicated Z-score or aortic root dissection; EL = ectopialentis; ELS = ectopialentis syndrome; FBN1 = fibrillin-1 mutation; FH = family history; MFS = Marfan syndrome; Syst = systemic score; US/LS = upper segment/lower segment ratio; Z = Z-score.

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