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CASE OF MARFAN'S SYNDROME PRESENTING A SPONTANEOUS PNEUMOTHORAX

Pulmonary Medicine

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ABSTRACT

A case of Right sided Spontaneous pneumothorax in a 12 year old girl with previously undiagnosed Marfan's syndrome. On physical examination she presented with marfanoid features.SP was managed with Intercostal chest tube drainage and Video assisted thoracic surgery. Marfan Syndrome itself and its respiratory manifestastions are not common , hence patients who are clinico-etiologically suspicious of spontaneous pneumothorax, marfan syndrome as a probable etiology should be kept in mind .

Background :-Spontaneous pneumothorax (SP) is a relatively rare condition in paediatric population with a bimodal peak age of occurance either in neonatal period or in late adolescence. In children SP is often caused by a tear in visceral pleura due to rupture of a subpleural bulla & often affects tall individuals with thin built1.

Marfan syndrome is a common inherited connective tissue disorder with typical skeletal, ocular and cardiovascular manifestations. Pulmonary involvement occurs less frequently, with SP being the most frequently reported (4–11%)2-4.

Marfan syndrome should be suspected in patients with SP and marfanoid habitus.

KEYWORDS

Spontaneous Pneumothorax , Marfan Syndrome

Case Report:- We report a 12 year old schoolgirl who presented with a 12 hour history of chest discomfort/pain and shortness of breath .At presentation she was dyspnoeic to talk and breath sounds were reduced on right side of chest. There was no history of previous trauma, smoking or any drug use or fever. Pneumothorax was managed with Conservative measures (Oxygen by Face Mask ,Rest) and insertion of a intercostal chest tube with underwater seal drain following which lung expanded &patient was relieved of symptoms. Patient was discharged after 8 days with removal of chest drain. Two months later she presented with Chest pain and shortness of breath with a recurrence of right sided pneumothorax following which patient underwent video assisted thoracoscopic surgery (VATS) and pleurodesis. No further recurrence was reported .Patient had many skeletal manifestations of marfan syndrome, Her height was 173 cm, arm span 192 cm ,metacarpal index of 8.6.A high arched palate was evident with arachnoidactyly and hyperextensible joints,both Wrist(Walker-Murdoch Sign) and Thumb(Steinberg) sign were positive. Bilateral superior subluxation of lens was present. Aortic root -33mm(at level of sinuses),Sino-tubular Junction -18.4mm,Assending Aorta 18.6mm,Aortic Arch 16.8 mm, Descending aorta 13.4 ,Main pulmonary artery 24mm dilated.

Discussion:- Marfan syndrome is a variable, autosomal dominant connective tissue disorder whose cardinal features affect cardiovascular system, eyes and skeleton. Additionally, other systems can be affected (lungs, skin and dura). Pulmonary manifestations include honeycombing, congenital bronchial malformations, bronchiectatic changes in the lower lobes, bullous emphysema and spontaneous pneumothorax.

Two studies published in 1980s and the Karpman et al[2] study in 2011 found a prevalence of pneumothorax in patients with Marfan syndrome between 4% and 11%, being most frequently described respiratory feature in those patients.

The increased risk of pneumothorax has been attributed to presence of apical blebs, bullae, abnormal connective tissue constituents in lung parenchyma or increased mechanical stresses in lung apices due to tall body habitus[2].

The causal gene for Marfan's syndrome, FBN1, encodes extracellular matrix glycoprotein fibrillin-1, which can be found in lung as a component of elastic fibres, it has been proposed that abnormalities of fibrillin result in connective tissue friability and laxity.

These features can result in chest wall deformity or flaccidity of small airways and terminal bronchioles, predisposing to premature airway closure, obstruction and air trapping leading to degenerative changes and emphysema, which apparently is main mechanism of pneumothorax in these individuals[2].

Karpman et al 2 suggested, not all individuals with Marfan syndrome have same increased risk of SP and use of available imaging studies such as CT scanning, may help physicians to classify patients into higher and low risk groups.

After initial diagnosis of Marfan syndrome, management by a multidisciplinary team is recommended. Patients should be referred to an ophthalmologist to exclude lens subluxation, refraction and visual correction, to an orthopaedic surgeon, for evaluation of skeletal manifestations that may require attention (eg, severe scoliosis, severe pectus excavatum). The existence of cardiovascular disease, should be evaluated by echocardiography, and should be referred to a cardiologist even in absence of severe disease,as β-blockers, are routinely prescribed.

Conclusion:- Clinical evolution of patients with Marfan syndrome is highly variable .Hence a high index of suspicion and profile of marfan syndrome, should should be kept in mind as diffrential diagnosis for marfan syndrome being a cause of SP.Over past three decades, there has been improvement in life expectancy of patients with Marfan syndrome which is probably due to advances in management of the condition.

References

1. O'Lone E, Elphick HE, Robinson PJ. Spontaneous pneumothorax in children: when is invasive treatment indicated? Pediatr Pulmonol 2008;43:41–6.
2. Karpman C, Aughenbaugh GL, Ryu JH. Pneumothorax and bullae in Marfan syndrome. Respiration 2011;82:219–24.
3 Hall JR, Pyeritz RE, Dudgeon DL, et al. Pneumothorax in the Marfan syndrome: prevalence and therapy. Ann Thorac Surg 1984;37:500–4.
4 Wood JR, Bellamy D, Child AH, et al. Pulmonary disease in patients with Marfan syndrome. Thorax 1984;39:780–4.