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CASE STUDY

MARFAN SYNDROME WITH PLEURAL EFFUSION: A CASE REPORT

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ABSTRACT

Marfan syndrome is a rare autosomal dominant disorder of the connective tissue, with skeletal, ligamentous, orooculofacial, pulmonary, neurological and the most fatal, cardiovascular manifestations. Pulmonary involvement occurs less frequently. We report a case of a 26-year-old male suffering from hours of sudden onset, progressive shortness of breath and right-sided chest pain. On physical examination, he presented decreased breath sound on the right side of chest, together with marfanoid habitus. The chest X ray revealed right sided pleural effusion and pleural fluid was drained by thoracocentesis. This case indicates that pulmonary symptoms like spontaneous pleural effusion, pneumothorax, emphysema can manifest as initial symptoms of undiagnosed Marfan syndrome.

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INTRODUCTION

Marfan syndrome is an autosomal dominant systemic disorder of connective tissue (Dean, 2002). Children affected by the Marfa syndrome carry a mutation in one of their two copies of the gene that encodes the connective tissue protein fibrillin-1(FBN 1) (von Kodolitsch, 2008). It has skeletal, ligamentous, orooculofacial, pulmonary, neurological and the most fatal, cardiovascular manifestations. The incidence of Marfan syndrome is approximately 2–3 in every 10,000 individuals, and pulmonary involvement occurs much less frequently. The diagnosis is commonly considered in a young person with a tall, thin body habitus, long limbs, arachnodactyly, pectus deformities, and sometimes scoliosis. Other clinical findings such as a high arched palate with dental crowding, skin striae, recurrent hernia or recurrent pneumothoraxmay increase suspicion. Family history may be helpful, but around 27% of cases arise from new mutation (Grimes *et al*., 2004). We here present a case with spontaneous pleural effusion as an initial diagnosis of Marfan syndrome.

Case report

A 26 years old male was admitted to emergency with hours of sudden onset, progressive shortness of breath and right sided chest pain.

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Chest X-ray was done which suggested pleural effusion in right lung (Figure 1). A chest tube was inserted into the thoracic cavity and the pleural fluid was drained. On physical examination he presented with decreased breath sounds on right side of chest, tall stature with lower segment greater than upper segment and long arm-span (Figure 2). Outward bowing of the legs was noticed along with dryscaly skin and long tapering fingers (Figure 3). Upper extremities showed long spidery fingers (arachnodactyly) with prominent finger joints (Figure 4).The chest was flat with prominent ribs (Figure 5) .Intraorally, high arched palate (Figure 6) was noticed along with the collapsed maxillary arch. Head and neck examination revealed a convex profile with long and narrow face. The supraorbital ridges were prominent (Figure 7). A special clinical test for evaluation of hyper extensibility included thumb (Steinberg) sign and wrist ((Walker) sign (Figure 8 and 9) which were both positive. On oculo-visual examination her unaided vision was 6/36 in each eye. Slit lamp examination was normal. The fundus was normal in both eyes and there were no signs of retinal degeneration. Objective refraction was -3.5/-1.5 /13 in right eye and - 4.75 /-2.00 /117 in left eye. With the subjective acceptance of-3.00/-1.00× 10° and -3.50 /-1.0× 120°, the visual acuity improved to 6/6 in each eye. With these findings he was provisionally diagnosed to have Mar fans syndrome with right sided pleural effusion.

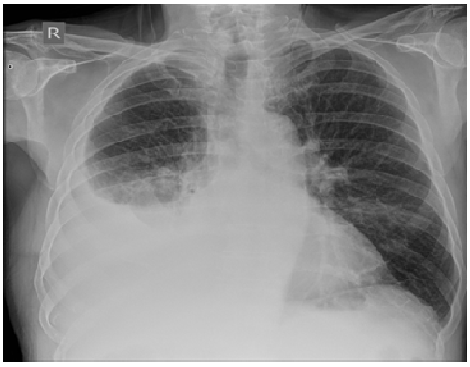

Figure 1. Chest X ray showing right sided pleural effusion

Figure 2. Tall stature with long Arm-span

Figure 3 Dry scaly skin with long tapering fingers

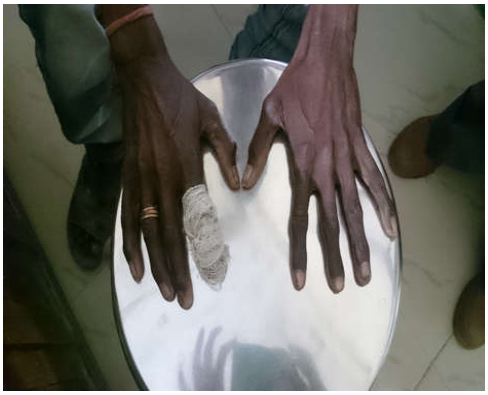

Figure 4 : Long spidery fingers (Arachnodactyly)

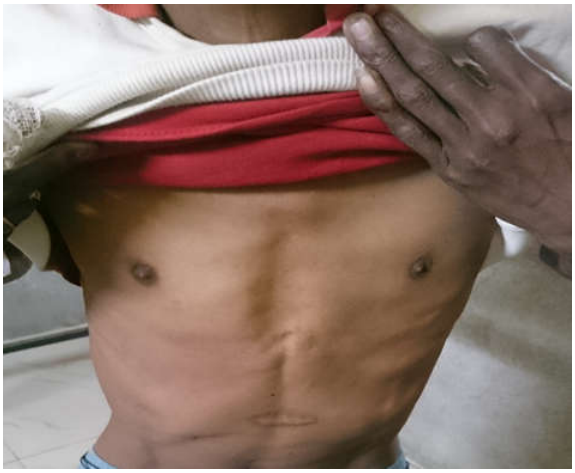

Figure 5. Flat chest with prominent ribs

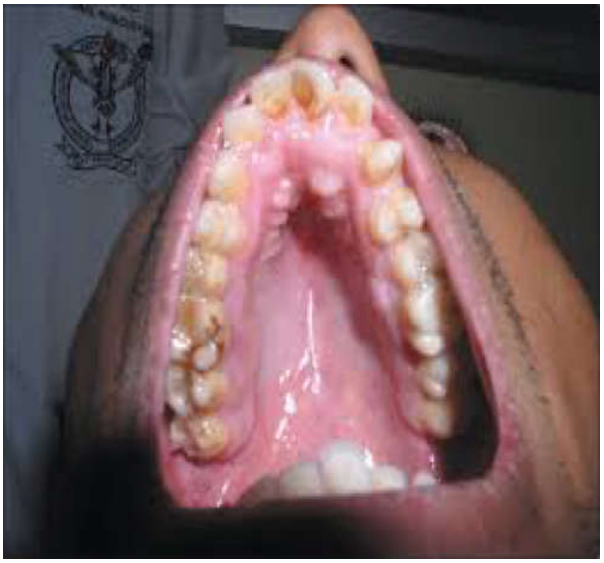

Figure 6. High arched palate

Figure 7. Convex profile with long narrow face

DISCUSSION

Fibrillin is an important component of the microfibrillar system that acts as a scaffold for elastogenisis. Classical Marfan syndrome is associated with a mutation in FBN1, the gene that encodes for fibrillin-1.

Figure 8. Positive thumb (Steinberg sign)

Figure 9. Positive wrist (Walker sign)

The pathophysiological outcomes of the degeneration of elastic fibers in Marfan syndrome seem to explain the majority of manifestations of this condition. Stiffness and reduced distensibility of the aorta in response to increased pulse pressure, is the main most important consequence of elastin degeneration (Dean, 2002). Recently, another hypothesis has emerged trying to explain the pathophysiology behind Marfan syndrome. Transforming growth factor β (TGFβ), a cytokine that regulates cell morphogenesis, is thought to contribute to the Marfan syndrome phenotype. Abnormal fibrillin causes failure of the sequestration of the inactive latent precursor of TGFβ, resulting in excessive TGFβ activation and thus producing the phenotypical manifestations of marfan syndrome (Lacro *et al*., 2007). Marfan syndrome primarily involves the skeletal, ocular and cardiovascular systems. Skeletal involvement is the most common clinical presentation; this is followed by cardiac problems thatoccur in 80-100% of cases; ocular presentation occurs in 60% of cases (Jones *et al*., 2007). Approximately, 16% of patients with Marfan syndrome have pulmonary symptoms (Wood *et al*., 1984), and pulmonary involvements may contribute to 10% of death in patients with Marfan syndrome (Chiu *et al*., 2014). Although pulmonary involvements are not generally considered a main feature of Marfan syndrome, many patients have a degree of underlying pulmonary pathology.

The pulmonary changes include widespread or patchy cystic changes, emphysema, and spontaneous pneumothorax; focal pneumonia or bronchiectasis, bullae, congenital pulmonary malformations (particularly middle lobe hypoplasia), and apical fibrosis (Dyhdalo, 2011). Skeletal manifestations are the cardinal signs of Marfa syndrome and usually gain the attention of a physician. The most common features include tall stature with the lower segment of the body greater than the upper segment and long, slender limbs, or dolichostenomelia; thin body habitus with increased arm span‐to‐height ratio; long, slender fingers, or arachnodactyly; deformities of the chest, such as pectuscarinatum or pectus excavatum, scoliosis and highly arched palate with crowded teeth and dental malocclusion. Otherless common manifestations include hypermobility of joints, flat foot (pes planus), reduced extension of elbows (<170°), and elongated face (dolichocephalia). Patients should be examinedfor arachnodactyly; positive wrist or Walker's sign (the distal phalange of the first and fifth fingers of the hand overlap when wrapped around the opposite wrist); and positive thumb or Steinberg sign (the thumb projects beyond theulnar border while completely opposed within the clenchedhand (Gray *et al*., 1998).

Ocular involvement is common and progres-sive. The more specific lesion for diagnosis is lens subluxation; however, one must also identify refrac-tion errors to preserve the maximum visual function (Paepe *et al*., 1996). It is well known that compound myopic astigmatism is the commonest refractive finding in Marfan's syndrome (Koenig, 1996; Paepe *et al*., 1996). Increasein axial length, flat corneal curvature andsubluxation of the lens, which causes the curvature of the lens to be steeper, maylead to these refractive findings (Jones *et al*., 2007). Marfan Syndrome is often highly lethal with life expectancy of about30-40 years. Cardiovascular complications are the major causes of mortality as was reported by Johnson *et al*. In infancy, death is usually from mitral regurgitation with or without tricuspid regurgitation. After infancy, death is usually from ascending aortic dissection and chronic aortic regurgitation (Johnson, 2012).

Differential diagnosis

Clinical diagnosis of Marfan syndrome is challenging because of the increased marfenoid features of other connective tissue diseases. Differential diagnosis could include homocystinuria, familial aortic dissection, familial arachnodactaly, Ehler Danlos syndrome and MEN IIb. Serum methionine must be carried out to rule out homocystinuria (Rangasetty *et al*., 2006). Molecular techniques have not been undertaken widely as a method to distinguish between Marfan syndrome and other similar-featured disorders, as it is not clear whether they can differentiate between those conditions with overlapping symptoms (Dean, 2002).

Conclusion

Marfan syndrome is an inheritable connective tissue disorder and is rare as compared to acquired connective tissue disorders. It is characterized by diverse clinical manifestations and the diagnosis is based on clinical criteria. Despite the morbidity and mortality associated with Marfan syndrome, appropriate medical and surgical management can improve and extend the lives of many patients, and advancing research holds the promise of further improvements in the future.

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