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MARFAN SYNDROME WITH PLEURAL EFFUSION

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ABSTRACT
Introduction: Marfan syndrome is rare autosomal disorder of the connective tissue affect both male and female equally, manifested with skeletal, ligamentous, orooculofacial, ophthalmic, pulmonary, neurological and most fatal cardiovascular manifestations. It share atypical features of others connective tissue diseases. Pulmonary involvement occurs less frequently. **Case Report:** we report a case of a 20 years old female with progressive shortness of breath over two weeks. on examination there was features suggestive of left side pleural effusion along with marfanoid habitus, chest x ray showed massive left side pleural effusion that drain by thoracocentesis. **Discussion:** Marfan syndrome is associated with a mutation in FBN1, the gene that encode for fibrillin-1, Fibrillin is an important component of microfibrillar system that act as a scaffold for elastogensis. **Conclusion:** This case illustrate that pulmonary symptoms like spontaneous pleural effusion, can manifest as initial symptom of undiagnosed Marfan syndrome.

KEYWORD: Marfan Syndrome, Pleural Effusion.

INTRODUCTION
Marfan's syndrome (MFS) is one of the most common autosomal dominant inherited disorders of connective tissue, affecting the heart (aortic aneurysm and dissection, mitral valve prolapse), eye (dislocated lenses, retinal detachment) and skeleton (tall, thin body build with long arms, legs and fingers; scoliosis and pectus deformity).

Clinically, two of three major systems must be affected, to avoid overdiagnosing the condition. Diagnosis may be confirmed by studying family linkage to the causative gene, or by demonstrating a mutation in the Marfan's syndrome gene (MFS1) for fibrillin (FBN-Í) on chromosome 15q21.

CASE REPORT
20 years old female not known to have any chronic illness, was admitted to Elmek Nimer university hospital complaining of progressive shortness of breath over 2 weeks, irritability and mild dry cough for 2 days, systemic review was unremarkable apart of anorexia and loss of weight.

On physical examination patient look ill irritable but conscious, cachexic, dyspneic, local chest examination on inspection the chest was flat with prominent ribs(fig. 5), bulging on left side which was move less, on palpation trachea and apex were shifting to the right with decrease chest expansion and tactile fremitus in the left, on percussion stony dullness was noted all over the left side, cardiovascular examination there was feature suggestive of mitral valve prolapse that confirmed later by Echocardigraphy, also patient was pale not jaundice, her palate was high arched (fig. 2) with long and narrow face (fig.4), tall stature, lower segment greater than upper segment and long arm span (fig. 1), long spidery fingers (arachnodactyly) with prominent finger and hands joints (fig. 3), the Steinberg and Walker signs were positive. slit lamp examination was normal in both eyes. Chest x ray was done that showed massive left side pleural effusion (fig. 6), diagnostic and therapeutic pleurocentesis was done. With all these finding she diagnosed to have Marfan's syndrome with massive left side pleural effusion.

Figure 1: Tall stature with long Arm-span.

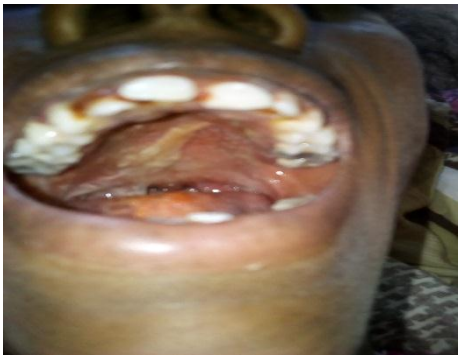

Figure 2: high arched palate.

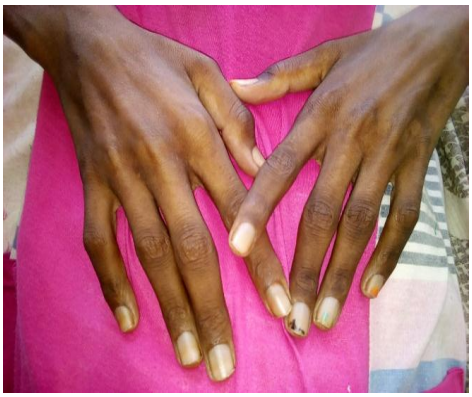

Figure 3: long spidery fingers (Arachnodactyly).

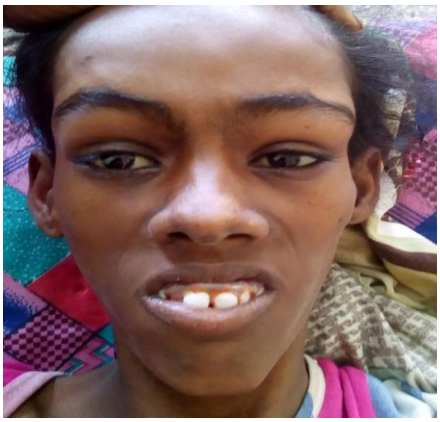

Figure 4: long narrow face.

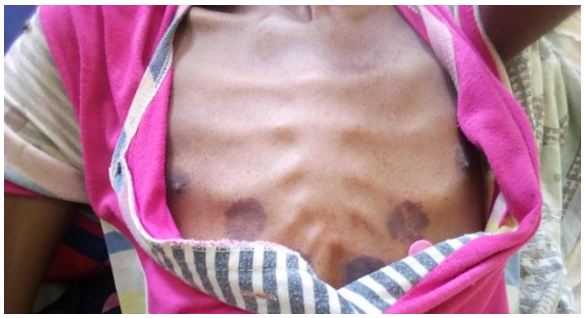

Figure 5: flat chest with prominent ribs.

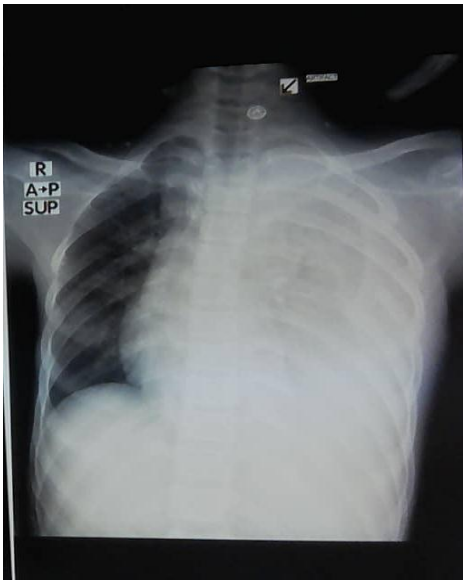

Figure 6: chest x ray showing left sided pleural effusion.

DISCUSSION

Fibrillin is an important component of microfibrillar system that act as a scaffold for elastogensis, classical marfan syndrome is associated with a mutation in FBN1, the gene that encode for fibrillin-1. MFS affects approximately 1 in 5000 of the population worldwide and 25% of patients are affected as a result of a new mutation. This group includes many of the more severely affected patients, with high cardiovascular risk. Other known associations with early death due to aortic aneurysm and dissection are: family history of early cardiac involvement; family history of dissection with an aortic root diameter of >5 cm; male sex; and extreme physical characteristics, including markedly excessive stature and widespread striae.

Histological examination of aortas often shows widespread medial degeneration, described as 'cystic medial necrosis'.

Chest X-ray is often normal but may show signs of aortic aneurysm and unfolding, or of widened mediastinum. Pneumothorax affects 11% and scoliosis is present in 70% of patients.

ECG may be misleadingly normal with an acute dissection. In conjunction with mitral valve prolapse, 40% of patients usually have arrhythmia, with premature ventricular and atrial arrhythmias.

Echocardiography shows mitral valve prolapse, and mitral regurgitation in the majority of patients. High quality serial echocardiogram measurements of aortic root diameter in the sinuses of Valsalva, at 90° to the direction of flow are the basis for medical and surgical management. CT or CMR can detect aortic dilatation and are useful in monitoring. The management depend on medical and surgical policy this include, Beta-blocker

therapy slows the rate of dilatation of the aortic root. ACE receptor blockers. In Marfan's there is up regulation of TNF-β, which is specifically inhibited by ACE blockers. Lifestyle alterations are required because of ocular, cardiac or skeletal involvement. Sports that necessitate prolonged exertion at maximum cardiac output, such as cross-country running, are to be avoided. Monitoring with yearly echocardiograms up to aortic root diameter of 4.5 cm, 6-monthly from 4.5 to 5 cm, and then referred directly to a surgeon who is experienced in aortic root replacement in Marfan's syndrome for elective surgery. Medical and surgical management have increased the overall survival rate. On average, 13 years of life is added when surgical survival is compared to that reported in the natural history of MFS. Genetic counselling: The condition is inherited in an autosomal dominant mode, with each child of one affected parent having a 50: 50 chance of inheriting the condition. Males and females are equallyoften affected. In 25% of all cases, the condition arises as the result of a spontaneous mutation in gene 5 of one of the parents. Fibrillin-1 gene mutations can be identified in 80% of those affected, confirming diagnosis and aiding prognosis.

Differential Diagnosis

Maranf syndrome it share atypical features of others connective tissues diseases so It is so difficult to differentiate and diagnosed marfan syndrome. Differential Diagnosis could include Ehler Danlos syndrome and MEN IIb. Homocystinuria must be rule out by measure Serum methionine.

CONCLUSION

Marfan syndrome is an inheritable connective tissue disorder and is rare as compare to acquired connective tissue disorder. It is characterized by diverse clinical manifestations and the diagnosis is based on clinical criteria. We report a case of a 20 years old female with progressive shortness of breath over two weeks chest x ray showed massive left side pleural effusion. This case illustrate that pulmonary symptoms like spontaneous pleural effusion, can manifest as symptom of Marfan syndrome.

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