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CASE REPORT ON MARFAN SYNDROME

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Abstract:

Marfan syndrome is a spectrum of disorder caused by a heritable genetic defect of connective tissue that has an autosomal dominant mode of transmission. The defect itself has been isolated to FBN1 gene on chromosome 15, which codes for connective tissue protein FIBRIN. A mutation result in an increase in protein called transforming growth factor ß. Abnormalities in this protein cause a myriad of distinct clinical problems, of which the musculoskeletal, cardiac and ocular system problems predominate. A case report on Marfan syndrome with mediastinal widening and sinus tachycardia was reported. This report underscores the significance of thorough family history and physical examination in the diagnosis of Marfan syndrome. Additionally, the effectiveness of therapy can be improved by having a proper insight about the pathology and clinical presentation of Marfan syndrome. Regular ECG, blood pressure monitoring and early initiation of beta blockers therapy as well as elective prophylactic surgical repair contribute to increasing the survival rate of Marfan patients.

Keywords: *Marfan syndrome, Mediastinal widening, Sinus tachycardia, Beta Blockers, Myopia*

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INTRODUCTION:

Marfan syndrome is an inherited disorder that affects connective tissue — the fibers that support and anchor your organs and other structures in your body. Marfan syndrome frequently affects the heart, eyes, blood vessels and skeleton. About 1 in 5000 people have marfan syndrome, including men and women of all race and ethnic groups. Some people can have marfan syndrome due to spontaneous mutation, and will be first to have this disease in their family[1]. There is a 50% chance of passing this mutation to progeny. People are born with marfan syndrome, but they may not notice any features until later in life. Its features include musculoskeletal:-long arms, legs and fingers, tall and thin body, curved spine, flat feet, chest sinks in or sticks out and crowded teeth. Cardiac: aortic root dilation, dissection. Eyes:-myopia, lens dislocation[2,3].

Diagnosis is based on examining both physical features and genetic/family history. The signs and symptoms are compared against ghent criteria. This diagnostic tool consists of major and minor criterias. Major criteria include enlarged aorta, dislocation of lens, family history, and musculoskeletal problems such as scoliosis (or flat feet). Minor criteria include myopia, long and thin face, loose joints, a high arched plate and unexplained stretch marks. Other diagnostic tool include echocardiogram and chest x-ray[4,5]. Standard medical therapy for marfan syndrome consist of ß –blockers which lowers heart rate, blood pressure and reducing wall of aorta. New treatment as a second option include ARB blocker-losartan which is an important alternative therapy to patient intolerant to ß –blocker. Aortic root surgery is also prefered.

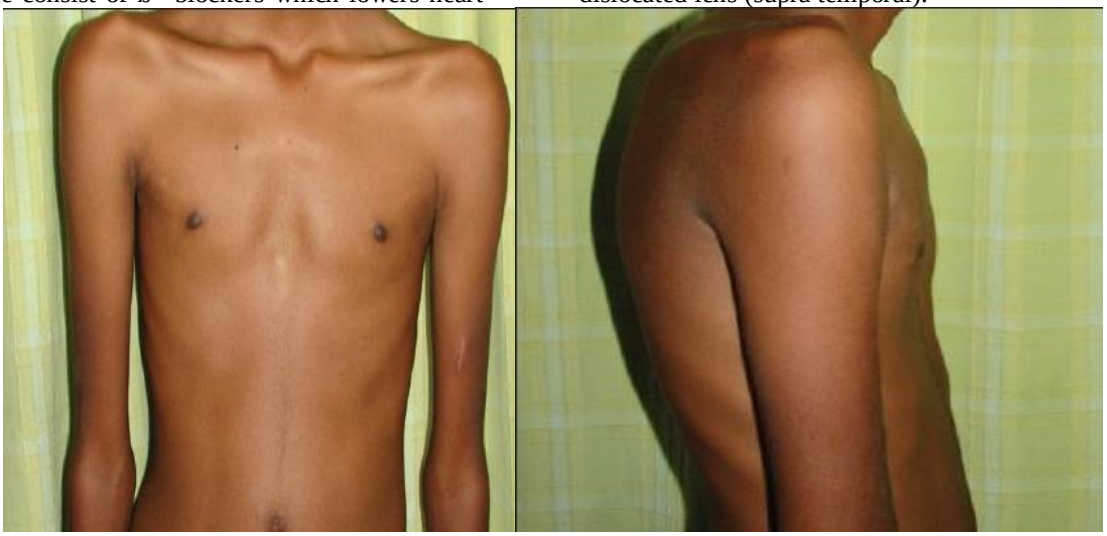
CASE REPORT:

A 21yr old young man was admitted in PMC hospital with complaints of tenderness in epigastric region, abdominal pain, vomiting and difficulty in swallowing. On preliminary examination itself the patient was found to have marfaroid features, long arms, legs and crowded teeth[6].

On investigating family history, the patient was found to be the tallest one of all the family members. No family history for marfan syndrome was found.

On general examination his B.P chart revealed increased systolic BP and Chest x-ray showed mediastinal widening and ECG shows sinus tachycardia. On final diagnosis patient was found to have acid peptic disease (upper GI endoscopy – ulceration) and marfan syndrome. He was treated with Syp.Sucralfate 10 ml, Inj.Pantoprazole 40 mg, T.Vitamin B 12 and T. Atenolol 25 mg.

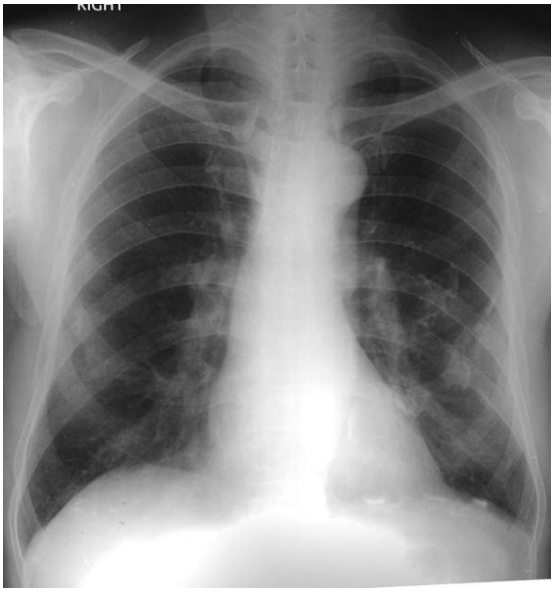
He was discharged after 6 days of hospital admission. After 3 weeks the patient consulted in the ophthalmology department of same hospital as he experienced some visual problems. On investigation he was found to have myopia or nearsightedness and dislocated lens (supra temporal).



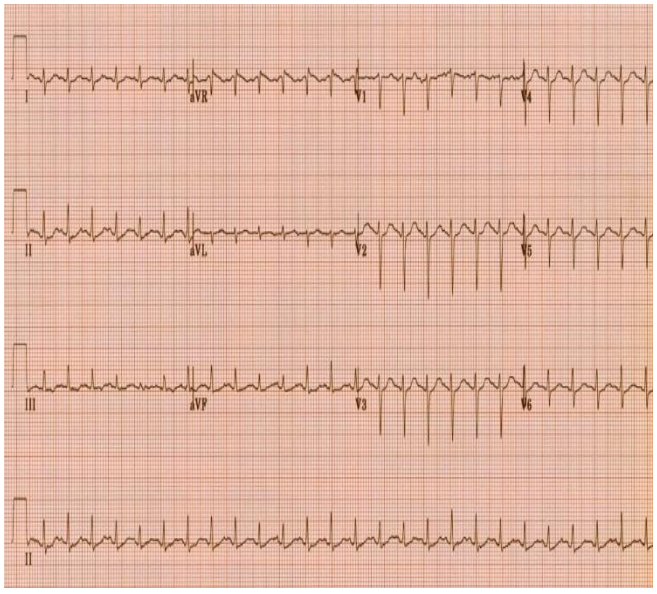
LAB PARAMETERS

PARAMETER	PATIENT VALUE
HAEMATOLOGY	
Hb	14.4
RBC	5.25 X 10^6
Plt	216 X 10^3
MCV	80.2
MCH	27.2
MCHC	34
BIOCHEMISTRY	
B.Urea	29
S.Creatinine	0.8
DIFFERENTIAL COUNT:	
%L	22.0%
%M	6.5%
%G	71.5%
ESR	30
URINE ANALYSIS:	
Sugar	Nil
Pus cells	2-4
Epithelial cells	1-2
LIVER FUNCTION TEST	
S.T Protein	7.6
S.T Albumin	4.8
S.T Globulin	2.8
S.T Bilirubin	0.7
SGPT	21
SGOT	17
S. Alkaline phosphatase	97

CHEST X-RAY



ECG



DISCUSSION:

Marfan syndrome is an autosomal inherited connective tissue disorder with diverse clinical manifestations. Although various studies have been conducted which aimed at improving the medical aspect of management, those trials produced conflicting results and generally involved relatively few patients.

In this report the patient received symptomatic treatment; Tablet Atenolol was given for treating mediastinal widening and increased systolic BP. For acid peptic disease he was managed with syrup Sucralfate and Injection pantoprazole. Laboratory investigation revealed elevated monocytes and ESR count but no treatment was provided. He presented with complaints of vomiting on admission but no anti-emetics were given. On discharge he was prescribed with Tablet Atenolol 25mg BD, Syrup sucralfate 10ml TID and tablet Pantoprazole 40mg OD and review after 1 week.

Three weeks later he experienced visual problems and consulted ophthalmology department of same hospital for further evaluation where he was found to have myopia and dislocated lens. Opthalmologist advised for eye glasses and periodic monitoring. Myopia and dislocated lens are the symptoms of disease progression of marfan syndrome.

CONCLUSION:

This report underscores the significance of thorough family history and physical examination in the diagnosis of Marfan syndrome. Additionally, the effectiveness of therapy can be improved by having a proper insight about the pathology and clinical presentation of Marfan syndrome. Regular ECG, blood pressure monitoring and early initiation of beta blockers therapy as well as elective prophylactic surgical repair contribute to increasing the survival rate of Marfan patients. Opthalmologic examination periodically to assess the disease progression is also necessary. New studies points out the treatment with second option includes ARB blocker-losartan as an important alternative therapy to patient intolerant to ß –blocker. Aortic root surgery is also preferred treatment for this.

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CONFLICT OF INTERESTS

The author declares no conflict of interest

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