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A case of Marfan syndrome with complications

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Abstract: *Marfansyndrome is a heritable connective tissue disorder inherited as an autosomal dominant trait with complete penetrance. There is involvement of cardiovascular, ocular, skeletal, pulmonarysystem, skin and dura.[1] There is mutations in FBN1 gene, which encodes large Glycoprotein, fibrillin.1.[2]*
Cardiovascular manifestations include valvular disease involving either mitral valve, aortic valve, or both. Aortic regurgitation can result from distortion of aortic valve cusps due to enlarged aortic root occurring in 15% to 44% of patients.
This is a case of 35 years female patient, multiparous, who presented with recurrent symptoms and signs of heart failure. She had history of high grade fever and joint pains. On examination, she had marfanoid features, signs of aortic and mitral regurgitation and pulmonary hypertension.She had aortic root aneurysm with dissection (Stanford type A) and also treated as a case of probable endocarditis. Early recognition of aortic aneurysm is very important to prevent progression to dissection in setting of Marfan syndrome to prevent complications.
Keywords*: Aortic, aneurysm, dissection, Marfan syndrome, multiparous, endocarditis*

I. Introduction

Marfansyndrome is inherited as an autosomal dominant trait with complete penetrance. The individuals present with involvement of cardiovascular, ocular, skeletal, pulmonarysystem, skin and dura.[1] Both medical and surgical treatment of aortic disease in these individuals has led to improvement of life expectancy.[3, 4] Cardiovascular manifestations include valvular disease involving either mitral valve, aortic valve, or both as present in this patient. Early recognition of aortic aneurysm is very important to prevent progression to dissection in setting of Marfan syndrome.

II. Case report

This is a case of 35years old female patient from Mbezi, Para 7 living 6, admitted with complaints of difficulty in breathing and swelling of both lower limbs for six months. The history of difficulty in breathing was of gradual onset, increasing in severity, initially on exertion, later even at rest accompanied with history of paroxysmal nocturnal dyspnea, palpitations and lower limb swelling suggestive of NYHA class IV heart failure. There was history of reduced urine frequency, high grade fever, non-migratory joint pains, cough, intermittent atypical chest pain and weight loss for more than a month. There was no history of sore throat, genital ulcers, hoarseness of voice or facial puffiness. During her course of illness, she had been admitted up to four times over the last 2 years due to heart failure and put on medications that included furosemide, spironolactone, isosorbidemononitrate and enalapril. She attended outpatient follow up irregularily. No history of tuberculosis, hypertension, diabetes mellitus or asthma was present. She reported to adhere well to her medications. On examination, she was fully conscious, febrile (38˚c) and moderately dyspneic. She had high arched palate, arm span:height 1.2, hyperextensible finger joints, arachnodactyl, swan neck deformity, finger and toe clubbing, pitting ankle, pedal and pretibial edema. There was noconjuctival paleness, oral thrush, peripheral or central cyanosis, tremors, skin lesions, thyroid enlargement, sternal tenderness, periorbital puffiness, palmar erythema or splinter hemorrhage. On cardiovascular examination, pulse rate was 100 beats per min, regular, of large volume and collapsing.. Blood pressure was 145/85mmHg with no significant difference between blood pressure of left and right arms. There was wide pulse pressure and positive Hill sign, Corrigans sign and Traube`s sign, pectusexcavatum, cardiomegaly, apical heave, left parasternal heave, loud P_2, apical pan systolic murmur radiating to axilla, early diastolic murmur at left sternal edge and no carotid bruit. Other systems examination finding included fine bilateral basal crepitations and tender hepatomegaly. Fundoscopy showed no evidence of papilloedema. No lens disclocation on slit-lamp. These above findings were suggestive of Marfan syndrome, signs of aortic and mitral regurgitation and pulmonary hypertension in heart failure. Blood work-up showed; WBC (K/UL): 15.59, Hb level (g/dl): 10.7, Platelets (K/UL): 199, Albumin (g/L): 35, Na (mmol/l): 142, K (mmol/l): 3.8, Creatinine (umol/l): 115, BUN (mmol/l): 10.1, ALT (U/L): 48, AST (U/L): 40 and ESR = 80mm/1st hour. Blood culture results showed no growth. Chest radiograph showed cardiomegaly and normal aortic knuckle. (Figure 1)

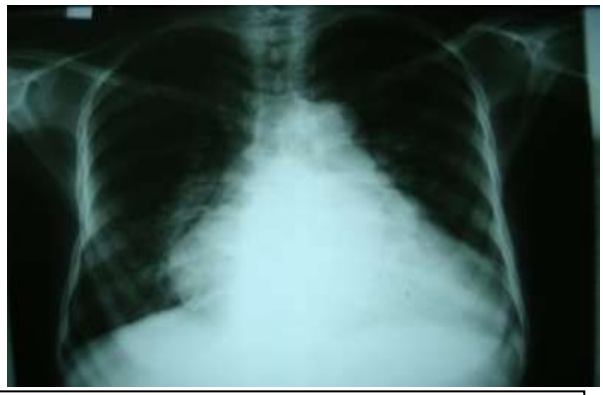

Figure 1: Chest radiograph showing cardiomegaly

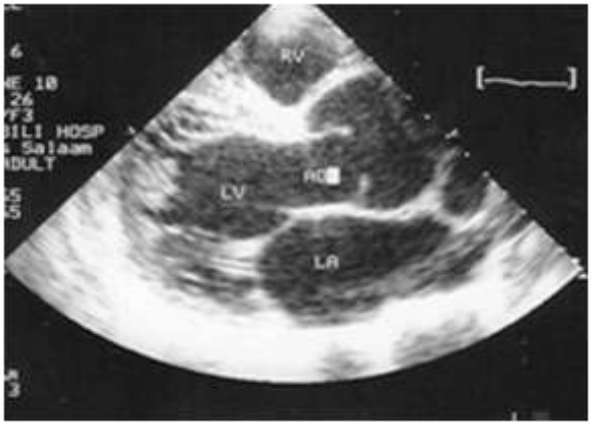

Figure 2: PLAX echocardiographic view showing dilated aortic root

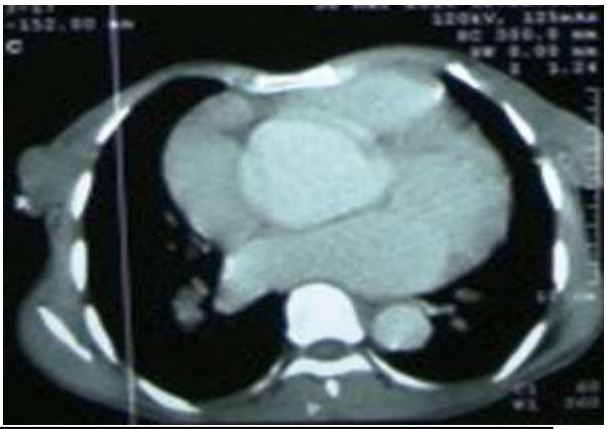

Figure 3: Chest CT scan showing intimal flap

Hand radiograph showed metacarpal index > 11. Electrocardiograph showed sinus tachycardia and left ventricular enlargement. Echocardiogram showed severe aortic regurgitation, mildfunctional mitral regurgitation and tricuspid regurgitation, dilated left ventricle, right atrium and inferior vena cave, dilated aortic root (aneurysm), pulmonary hypertension and ejection fraction was 53%. (Figure 2) Chest CT scan showed dilated aortic root of 7.1cm, left ventricular enlargement and intimalflap.(Figure 3)

In the ward, she had clinical improvement including reduced difficulty in breathing, fever subsided and NYHA class improved. Her medications were intravenous furosemide, intravenous ampicillin and gentanycin, tablets spironolactone, captopril and isosorbidemononitrate. Metoprolol tablets was also added. She received the intravenous antibiotics for 6 weeks. Her hemodynamics improved significantly. She was then planned for referral for aortic root surgery.

III. Discussion

Marfansyndrome is a heritable connective tissue disorder having prevalenceof 1 in 5000 individuals. It is inherited as anautosomal dominant trait with complete penetrance. The individuals present with involvement of cardiovascular, ocular, skeletal, pulmonarysystem, skin and dura.[1]The condition is due to mutations in FBN1 gene, which encodes a largeGlycoprotein, fibrillin.1.[2]

Both medical and surgical treatmentof aortic disease in these individuals with Marfan syndrome has lead to improvement of life expectancy.[3, 4]

Cardiovascular manifestations include valvular disease involving either mitral valve, aortic valve,or bothvalves as present in this patient. Mitral valve prolapse is the most prevalent cardiovascular manifestation affecting more than a third.[5]This patient had aortic regurgitation. Aortic regurgitation canresult from distortion of aorticvalve cusps due to enlarged aortic rootand this occurs in 15% to 44% of patients.

Literature shows it is generally recommended to do prophylactic aorticroot replacement with or without valve sparing in these patients at aortic size of at least5.0 cm.[6, 7]The risk for dissection or rupture is such that at aneurysm size of at least6 cm, there is up to a 4-fold increase incumulative risk of aortic rupture ordissection as occurred in this patient.[6]Our patient was multiparous. It has also been found that women with Marfan syndrome are at a very high riskfor aneurysm and dissection during pregnancy/ or subsequent pregnancies andshould be counseled before pregnancy about the high risk. Though, in pregnancy the risk for dissection islow if aortic root diameter is less than 4.0cm.[8]Review of literature shows that patients with Marfan syndrome have dilatation of ascending aorta including root and, dissections of aorta mainly in second and third trimesters.[9]Overall, the risk factors for aortic dissection include; aortic diameter greater than 5 cm, aortic aneurysm extending beyond sinus of valsalva, rapid rate of dilatation (1.5 mm per year in adults) and a positive family history.

Infective endocarditis is an infection that occurs and, in general population, it has an estimated annual incidence of 3 to 9 cases per 100,000persons in industrialized countries.[10]Our patient was also managed as a case of probable infective endocarditis due to high grade fever. There have been reports of endocarditis occurring in marfanoid patients with musculoskeletal and cardiovascular features including severe aortic regurgitation or mitral regurgitation.[11, 12]

Marfan syndrome can be complicated with occurrence of ascending aortic dissection and infective endocarditis. This poses a great challenge in the management of the patient due to high rate of mortality especially in resource limited settings in terms of access to surgical interventions.

IV. Conclusion

Cardiovascular manifestations in Marfan syndrome include valvular disease involving either mitral valve, aortic valve, or both. This case is presented because early recognition of aortic aneurysm is very important to prevent progression to dissection especially in setting of Marfan syndrome and multiparity complicated with infective endocarditis.

Consent: Informed consent was obtained from the patient
Conflict of interest: None

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