

Rheumatic heart disease in a patient with Marfan's syndrome

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ABSTRACT

Marfan's syndrome (MFS) is an inheritable autosomal dominant disorder affecting the connective tissue with defect isolated to FBN1 gene on chromosome 15, which codes for the connective tissue protein fibrillin. The malfunctioning of the above gene has been implicated with predominantly heart, musculoskeletal and ocular problems. The main cardiovascular manifestations in Marfan's syndrome are mitral valve prolapse and aortic dilation which according to literature database have a prevalence of 76% and 62% respectively in adult population with Marfan's syndrome. The case reported is that of a patient who presented with features of congestive heart failure secondary to rheumatic heart disease who also has clinical and echocardiographic features of Marfan's syndrome. This report highlights the coexistence of two major cardiovascular abnormalities, one inherited and the other one acquired which had not been previously

reported to the best of our knowledge so far in Nigeria and indeed West Africa.

Keywords: Aortic dilatation, Marfan's syndrome, Mitral valve prolapse, Rheumatic heart disease

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INTRODUCTION

Marfan syndrome is a multi-systemic connective tissue disorder usually associated with mutation in the gene which encodes for the protein fibrillin, and occasionally with mutation in TGFBR1 or 2. About 27% of cases arise from a new mutation. Marfan syndrome is a variable autosomal dominant disorder with characteristic cardiovascular, eye and musculoskeletal features. Significant morbidity and mortality has been associated with progressive aortic dilatation, aortic valve incompetence which may lead to aortic dissection or rupture. Although, mitral valve prolapse, lens dislocation, myopia, and arthritis have also been implicated in causing substantial morbidity [1].

Clinical suspicion is often entertained in young individuals with tall, lean body habitus, arachnodactyly and pectus deformities. Sometimes, recurrent pneumothorax may increase its suspicion. The

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occurrence of a positive family history may be helpful, as about three-quarter of patients will have a family history of the syndrome [2]. To make the diagnosis of Marfan syndrome more consistent and of more prognostic value, the Berlin diagnostic criteria of 1988 [3] were revised and the clinical features codified as the Ghent nosology in 1996 [4]. Prophylactic medical treatment to protect the aorta with regular follow-up helps prevent or delay serious complications [2].

Although rheumatic heart diseases are nearly forgotten in developed countries, it is the commonest cardiovascular disease of children and young people in developing countries. Most patients initially present with shortness of breath and, without appropriate intervention, may progress to heart failure. RHD should be strongly considered a differential, in patients of Sub-Sahara African descent with a valvular murmur [5, 6].

CASE REPORT

A 25-year old male who presented at the medical out-patient unit of our hospital with progressive breathlessness of three months duration, initially on moderate exertion then progressing to occur at rest. He had orthopnoea, paroxysmal nocturnal dyspnoea, non-productive cough, right upper abdominal swelling and easy fatigability. He didn't give history of recurrent sore throat in the past or history suggestive of acute rheumatic fever. There was a family history of similar physical habitus in his father and two older siblings. His father died few years earlier while being managed for congestive cardiac failure in another hospital. On general physical examination, he was found to have marfanoid habitus with reduced upper/ lower segment ratio less than 0.85, arm span to height ratio greater than 1.05, presence of wrist and thumb [Steinberg] sign, arachnodactyly, clinodactyly, pectus carinatum, and high arched palate as shown in Figure 1. On cardiovascular examination, the pulse rate was 102 beats per minutes, regular, small volume, he had thickened arterial wall and locomotor brachialis, blood pressure was 100/60 mmHg, supine jugular veins were engorged, apex beat at the eighth left intercostal space anterior axillary line, was heaving. There was also left parasternal heave, soft first heart sound, second heart sound with louder pulmonary component, had apical pansystolic murmur that radiated anteriorly and to the axilla, along with a diastolic murmur that was best heard at the left sternal border.

On respiratory examination, he was tachypneic, with dull percussion note over the right lower lung zone laterally and posteriorly, the breath sound was vesicular with coarse crepitations in the right lower lung zone laterally and posteriorly with basal fine crepitations on the left. Abdominal examination revealed a distended abdomen with right upper abdominal region prominence, he had

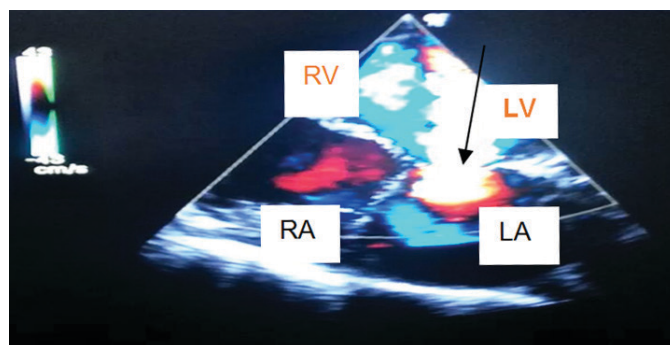
right indirect inguinal hernia which was reducible, liver was 6cm distended below the right costal margin with a span of 16cm and there was no ascites. Central nervous system examination was normal. Ocular examination didn't reveal presence of ectopia lentis or other ocular disorder.

Routine blood investigations were within normal limit. Chest X-ray showed features suggestive of pulmonary edema and massive cardiomegaly. Electrocardiogram showed sinus tachycardia, left ventricular hypertrophy and left atrial enlargement. Echocardiographic examination showed mitral valvular orifice area (MVOA) – 1.2 cm², mitral valvular gradient (MVG) – 34/22 mmHg, Pulmonary artery systolic pressure (PASP) – 58 mm of Hg with fusion of the anterior mitral commissure leaflet, he had severe dilatation of the aortic root (7.8cm) and effacement of the sino-tubular junction with severe aortic and mitral valve regurgitation as shown in Figures 2 and 3. There was no left atrial appendage clot. He was placed on anti-failure regimen including low dose losartan, intravenous antibiotics and anticoagulant. He is being followed up in the cardiology clinic.



LA: Left atrium, LV: Left ventricle, AOD: Aortic root dimension, RV: Right ventricle

Figure 1: 2-D echocardiography image (parasternal long axis view) showing massive aortic root dilatation, densely sclerosed mitral valve leaflets, cardiac chamber dilatation with spontaneous echo contrast in the left atrium, left ventricle and ascending aorta.



LA: Left atrium, LV: Left ventricle, RA: Right atrium, RV: Right ventricle, Black arrow: Aortic regurgitation jet

Figure 2: 2-D echocardiography image, (apical 5 chamber view) showing severe aortic regurgitation.

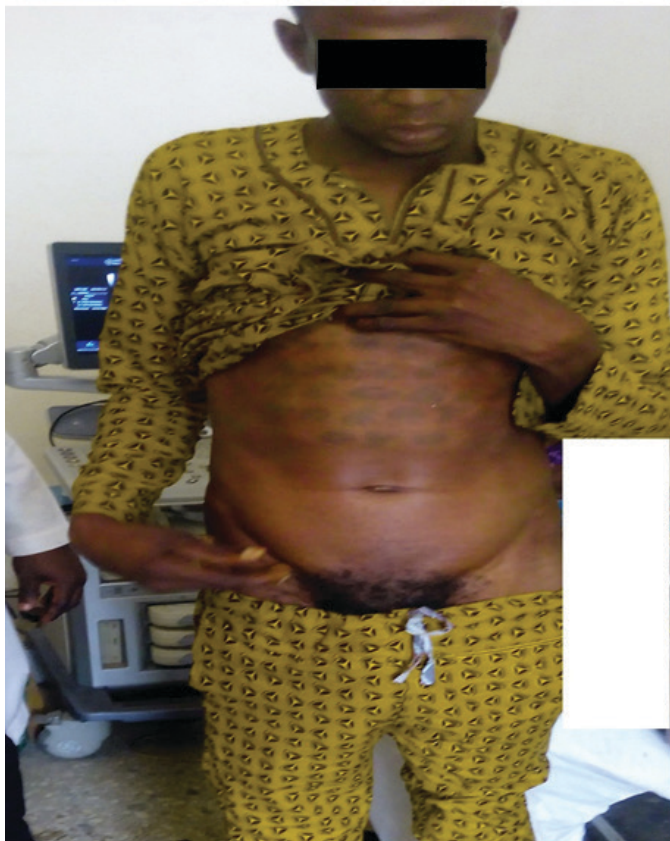


Figure 3: Marfanoid habitus with reduced upper/ lower segment ratio less than 0.85, arm span to height ratio greater than 1.05, presence of wrist and thumb [Steinberg] sign, arachnodactyly, clinodactyly and pectus carinatum were observed.

DISCUSSION

Marfan syndrome is a heritable autosomal-dominant disorder caused by mutations in one of the genes for fibrillin-1, which is a structural protein that is the major component of microfibrils of elastin. The progressive dilatation of the aorta has been attributed to the defect in elastin activity and also the reduced quantity in connective tissues due to the mutation [7]. The most common cardiovascular diseases reported in MFS are mitral valve prolapse (MVP) and aortic dilatation. MVP was reported in a series to be present in around 75% of cases, with increased prevalence in adolescence [8, 9]. MFS patients are said to be at increased risk of sudden death due to aortic dissection and rupture with a reduced mean life expectancy estimated at about 32 years in a study. Heart failure from valvular regurgitation sums up to the death toll [8]. Some studies however, showed biventricular and atrial systolic and diastolic dysfunction in a series of adult patients with MFS without significant valvular disease [10–13].

Our patient had RHD with moderate mitral stenosis and mitral regurgitation and severe aortic root dilatation with a risk of aortic dissection, rupture and family history

of sudden death. Treatment of this condition is often very complex as progressive valvular disorder which may require surgical intervention may be associated with a very high surgical risk which is even very expensive and not within the reach of the patient. Regular antibiotic prophylaxis, anti-failure regimen, anticoagulants to prevent cardio embolic stroke, low dose angiotensin receptor blockers and beta blockers with regular clinic and echocardiographic follow up constitute the current options of management adopted in this patient.

No specific laboratory investigation is required to make the diagnosis of Marfan's syndrome but genetic testing can be done to confirm it. Even though valvular abnormalities are common in Marfan syndrome, rheumatic valvular heart diseases are rarely reported even from Africa. No specific surgical procedure exist to cure Marfan's syndrome. There is evidence to show that angiotensin receptor blockers such as losartan may have potential effect to reverse some of the clinical manifestations of MFS such as aortic root dilatation, mitral valve prolapse or skeletal muscle dysfunction [14]. Medical therapy in the management of MFS is targeted at reducing cardiovascular decompensation. Medical therapy include use of beta blockers and afterload reducing agents which may reduce or halt the rate of progression of aortic root dilatation and dissection, reduction of aortic stiffness and improved cardiac compliance. Other medical therapy that can be used include nitroprusside, calcium channel blockers, e.g., verapamil [15, 16]. Non-operative methods such as bracing or thoracolumbosacral orthosis can be used to correct the scoliosis [17]. Surgical therapy options available for this patient include mitral valve repair and aortic root repair with replacement of the valves [18]. Percutaneous mitral balloon valvotomy and percutaneous transmitral balloon commissurotomy had been successfully done for patients with severe mitral valve stenosis and Marfan's syndrome in some case reports [19, 20].

CONCLUSION

This report highlights the rare coexistence of two major cardiovascular abnormalities, one inherited and the other one acquired. This is associated with significant cardiovascular risk and morbidity. Early screening and diagnosis are important measures to reduce the increased cardiovascular risk associated with these conditions especially in Africa where facilities for surgical interventions are not very available.

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successfully treated with percutaneous transmitral balloon commissurotomy – Report of first case. *J Cardiovasc Dis Res* 2013;4(4):257–9.

Author Contributions

Akinlade Olawale Mathias – Conception of the work, Design of the work, Acquisition of data, Analysis of data, Interpretation of data, Drafting the work, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Yusuf Abiola M. – Interpretation of data, Drafting the work, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Akintunde Adeseye Abiodun – Conception of the work, Design of the work, Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

Opadijo Oladimeji George – Interpretation of data, Revising the work critically for important intellectual content, Final approval of the version to be published, Agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved

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Written informed consent was obtained from the patient for publication of this article.

Conflict of Interest

Authors declare no conflict of interest.

Data Availability

All relevant data are within the paper and its Supporting Information files.

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