ORIGINAL RESEARCH PAPER

General Medicine

CASE STUDY- 25 YEAR OLD FEMALE MARFAN PRESENTING WITH AORTIC ROOT ANEURYSM:OPERATED BY DAVID PROCEDURE

KEY WORDS:

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INTRODUCTION

The incidence of MFS is among the highest of any heritable disorder: about 1 in 3000/5000 births in most racial and ethnic groups. Most of the patients with typical Marfan phenotype harbor mutations involving the gene (FBN1) encoding the connective tissue protein fibrillin -1.

Mutations are generally inherited as autosomal dominant traits, but about one-fourth of patients have sporadic new mutations.

In individuals with atypical presentations reminiscent of MFS, an inactivating mutation in a gene encoding for transforming growth factor-beta receptor (TGFBR) may be responsible.

FBN1 is a large gene (65 exons) located at chromosome 15q21.1. The fibrillin-1 protein contains many cysteine-rich domains homologous to those observed in epidermal growth factor (EGF) and the latent transforming growth factor beta binding proteins (LTBPs). Fibrillin-1 is an important matrix component of both elastic and nonelastic tissues. It is the main constituent protein of extracellular microfibrils that are thought to contribute to the formation and maintenance of elastic fibers.

Mutations in TGF-beta receptor 2 (TGFBR2) and TGFBR1 genes have been linked up to 10 percent of cases with the Marfan phenotype.

MFS was initially characterized by a triad of features:

1. skeletal changes that include long, thin extremities, frequently associated with loose joints
2. reduced vision as the result of dislocations of the lenses (ectopia lentis)
3. aortic aneurysms

Patients with structurally normal aortic valve leaflets and those Aortic Regurgitation is secondary to dilatation of Sino tubular junction or aortic annulus , may be undergo a valve sparing root replacement-Reimplanting the native valve within a Dacron graft ;David procedure.

Presentation

25 years old housewife, presenting with palpitations since 6 years . she was apparently alright 6 yrs ago, then she started experiencing palpitations in initial 3 years ,which was insidious in onset and gradually progressive in nature. Initially occurred on exertion and used to get relieved on rest. Then she complained of palpitations even on rest and doing her daily routine chores .No history of dyspnea, angina orthopnea, pnd,hemoptysis. No history of lower limb swelling /abdominal distension / decreased urine output / blackening of fingers. No history of joint pain / sore throat /prolonged fever / skin lesions /involuntary movements.No other known comorbidities .She has been following up with a doctor near to her home and on her last visit (i. e 6 months back.)she was advised to get admitted under tertiary care.

On examination patient is tall stature with increased length of long bones, thin built and poorly nourished , Marfanoid features are observed in this patient , like-down sloping palpebral fissure and high arched palate ,slender fingers, arachnodactyly .Arm span >> Height (arm span-176.53 cm height -163cm A/ H -1.083 { normally A/H >1.05 })

Decreased ratio of upper segment and lower segment (upper segment -57cm lower segment-106 cm ; US / LS-0.56 { normally ratio range 0.78-0.85}),Walker- Murdoch's sign.

SCORING OF SYSTEMIC FEATURES-Wrist &Hand sign - positive-3 points

Reduced upper segment/lower segment & increased ARM span- 1 point
Scoliosis -1 point
Facial features-1 point
Myopia-1 point
Systemic score -7 points

So according to Revised Ghent Criteria- diagnosis of Marfan syndrome

As, isolated patient (no family history) with Aortic Root Dilatation (Z>=2) and systemic score of >=7

Vitals -
Pulse 96 bpm regular rhythm , high volume ,rapid rising collapsing pulse. All peripheral pulses were well felt. No radio -radial delay or radio-femoral delay.
BP- right upper limb- 120/60 mmHg
Left upper limb -110/60 mmHg
Right lower limb -130/60 mmHg
RR-22/min abdominothoracic regular
Normal JVP

Systemic Examination

CVS- INSPECTION-Mild precordial bulge is present,apex impulse seen in 6th ICS at MCL, supra stern also pulsations visible.

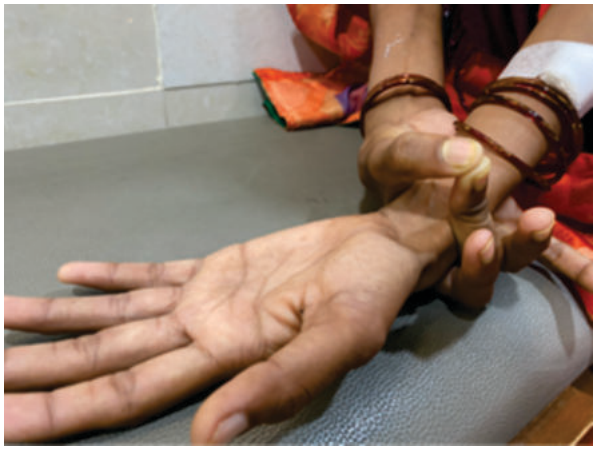
PALPATION-Inspectory findings are confirmed, apical impulse palpated in 6th left ICS 1cm lateral to MCL

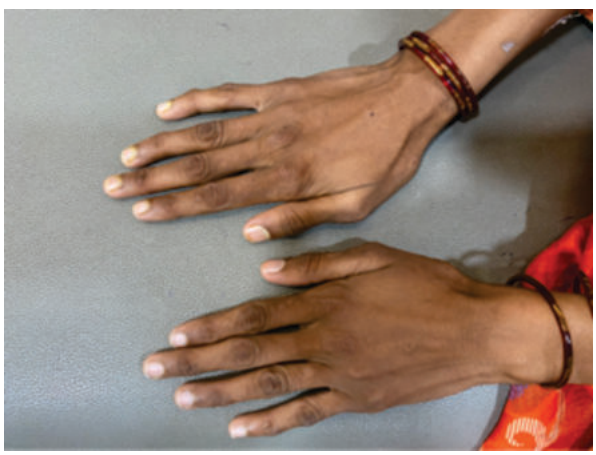
PERCUSSION-Liver dullness in 5th ICS, Left heart border corresponds to apex and Right heart border is retro stern alert

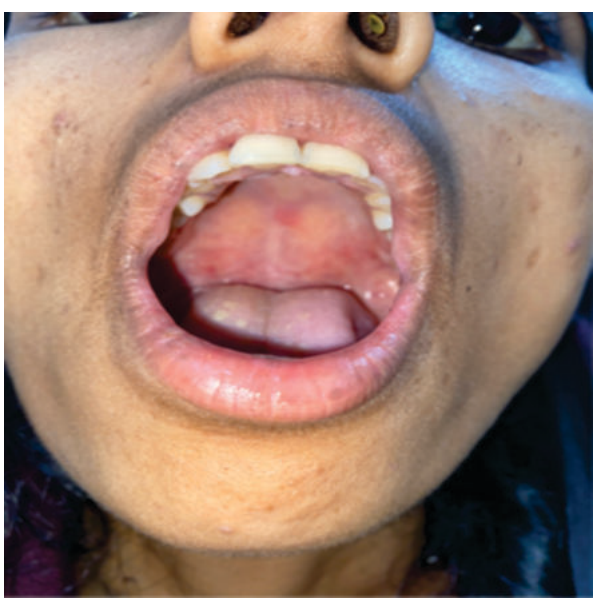
ASCULTATION- S1 normal, S2 normal split with no S3 /S4
High pitched blowing early diastolic murmur at left 3rd ICS long murmur with duration > 2/3rd of diastole best heard with diaphragm of stethoscope, with breath held in expiration and leaning forward.

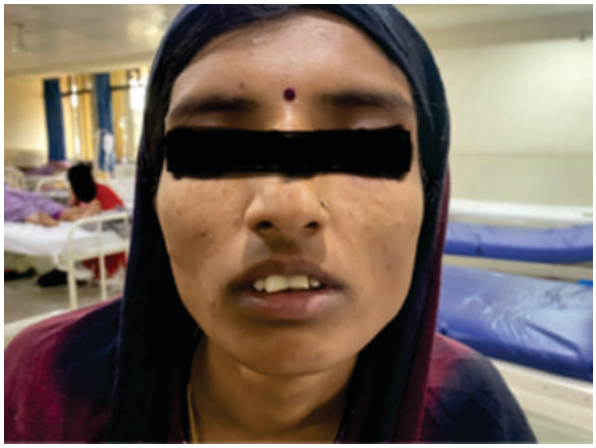
Short MDM at apex heard with bell of stethoscope low pitched with no pre systolic accentuating.

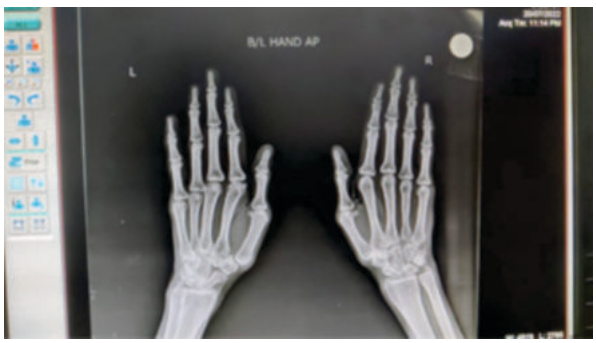
OTHER SYSTEMS- all normal.





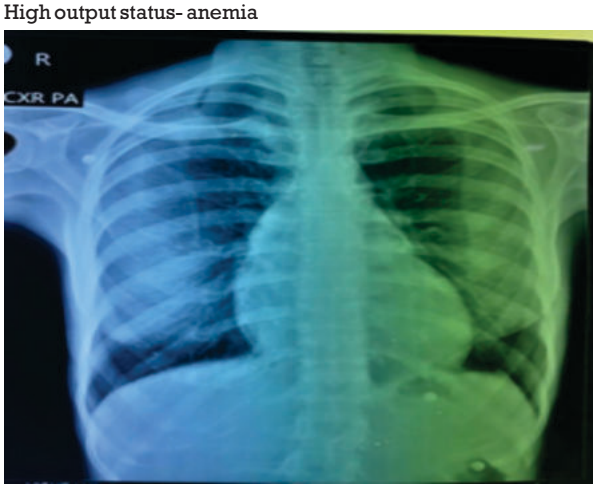


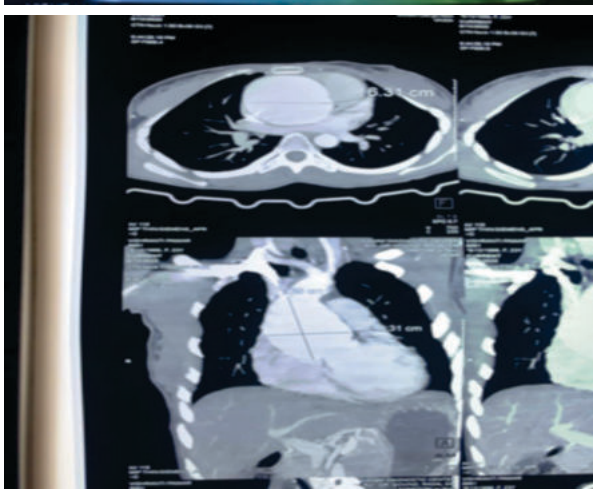


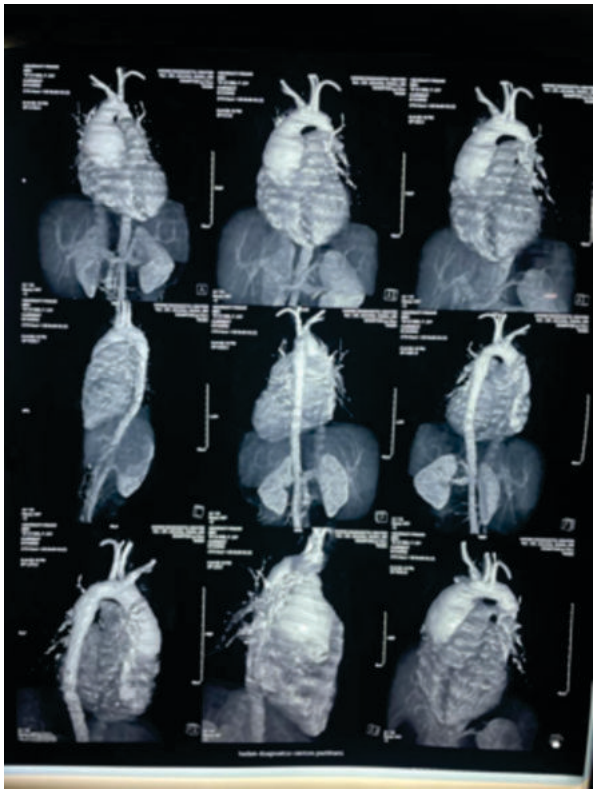


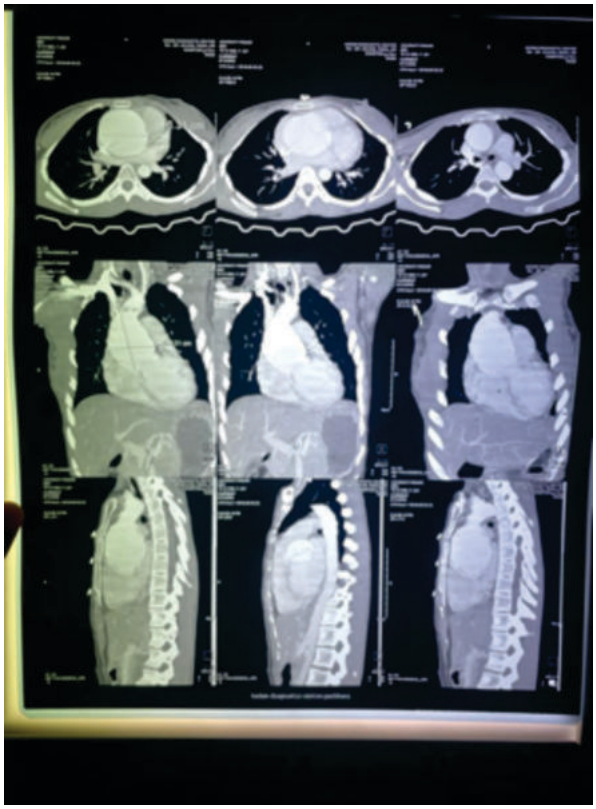
Differentials

Cardiac- mitral valve prolapse, valvular heart disease , cardiomyopathy
Endocrine-hyperthyroidism
High output status- anemia









Note; The aortic root dilatation was repaired by DAVID PROCEDURE.

INVESTIGATIONS

Pre - Operative-

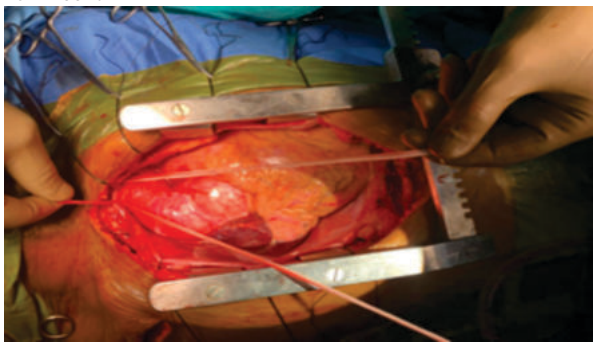
1. Transesophageal echocardiography -s/o-gross aneurysm of the aortic sinuses and ascending aorta (measuring-58.7mm) with non coaptation of the aortic valve causing diastolic orifice. dilated lv , LVEF reduced – 45-50%.
2. Ct aortogram- s/o – extensive aortic root dilatation measuring 6.3 cm regarding its maximum dimensions as well as the sino tubular junction involve segment measures 7 cm in length.

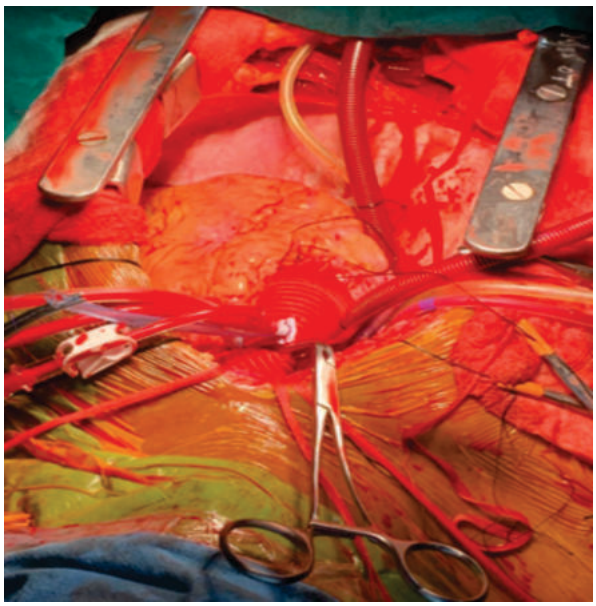
POST -David's Procedure-

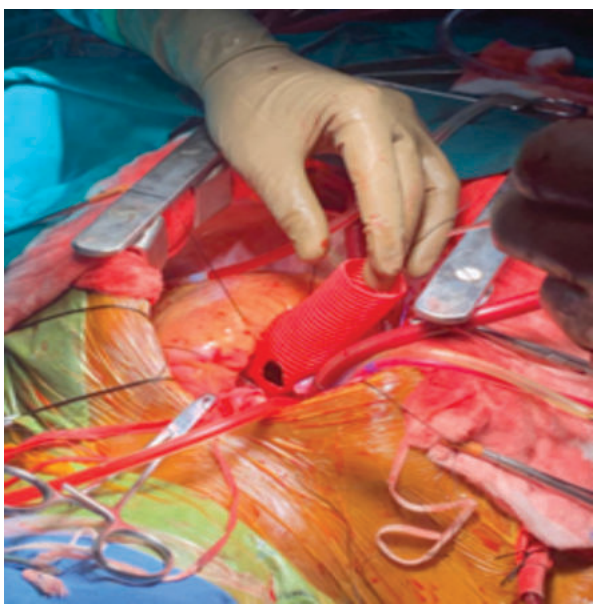
ECHOCARDIOGRAPHY-s/o post aortic graft,Trivial AR, Trivial TR-31mmHg,No AS/MS/MR

Norma cardiac chambers.

Normal Left ventricle cavity size with normal LV function LVEF-60%







Diagnosis

Chronic valvular heart disease in the form of moderate to severe AR with etiology being Aortic Aneurysm with Marfans syndrome in sinus rhythm not in CCF/ Infective endocarditis.

DISCUSSION

Marfan is an inherited connective tissue disorder with defect of microfibrillar component protein-fibrillin -1 gene located at chromosome 15q. In addition to directing elastogenesis and providing structural support to the tissues, fibrillin -1 interacts with latent transforming growth factor beta (TGF-beta) – binding proteins and controls the activation and signaling of TGF-beta .The abnormal fibrillin-1 in MFS leads to excess free TGF-beta , which promotes aortic disease. Multisystem is affected ,like skeletal ,cardiovascular system & ocular. Skeletal system involves tall stature , dolichostenomelia , arachnodactyly with hypermobility, dolicicephaly , scoliosis. Cardiovascular system -mitral valve prolapse , aortic aneurysm ,aortic dissection and aortic rupture. Ocular -ectopia lentis, early myopia, flat cornea and hypoplastic iris .Aortic dilatation in MFS affects most prominently the sinuses of Valsalva but distal aortic aneurysms and dissections may occur.

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