A Case Of Marfan Syndrome With Ascending And Arch Of Aorta Aneurysm Presenting With Type A-Dissection Of Aorta.

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Introduction:

- First described by Antoine – Bernard Marfan in an 1896 case report of a young girl with unusual musculoskeletal features .
- Hereditary disease which has AD inheritance because of mutation in the *fibrillin-1 gene-chromosome 15*.
- It affects *connective tissue* of the body- dolichostenomelia, mainly involves
  - ***CARDIOVASCULAR SYSTEM*** : *Ascending aorta aneurysm -annuloaortic ectasia with high risk of dissection (root diameter of > 4.5cm) , mitral valve prolapse , aortic regurgitation secondary to root dilatation*.
  - **OCULAR** : Bilateral ectopia lentis (40 – 56 %), myopia (28%) and retinal detachment (0.78%).
  - **SKELETAL SYSTEM**: Scoliosis, pectus excavatum, pectus carinatum, positive thumb & wrist sign.
- It is usually diagnosed with *2010 Revised Ghent Nosology with a score of more than 7*.
- Marfan syndrome incidence of acute aortic dissection is 1/10,000(0.01%).
- When the aortic root is dilated more than 5cm, it is usually managed surgically by *Bentall procedure /David valve sparing reimplantation operation* depending on involvement of aortic valve.

CASE REPORT:

- Female / 40 yr came with tearing type retrosternal chest pain radiating to neck and back of chest, and breathlessness of NYHA class 3 .
- On examination: conscious and coherent

  HR = 78/min, regular, all peripheral pulse felt ,

  BP = 150/60 mm of Hg both upper limb,

  180/50mm of Hg , lower limbs

  Lumbar scoliosis, pectus carinatum, upper segment to lower segment ratio 0.71 ,

  Increased arm span to height ratio with positive thumb sign.
- Cardiovascular examination:

  loud ,long EDM heard at 3rd left ICS, ESM grade 3/6 at Aortic area and MDM at apex.
- ECG : Sinus Rhythm, LVH with volume overload.
- Chest x ray: LV type apex ,with unfolding of arch of aorta with lumbar scoliosis .
- Echo : Aortic root of 4.78cm(at the sinuses of Valsalva) with severe AR with dissection flaps in the root of aorta, good LV funtion.
- TEE : Aortic root aneurysm with dissection of aorta type-A with dissecting flaps
- Referred to CT surgery department for BENTALL procedure.

Figure .1

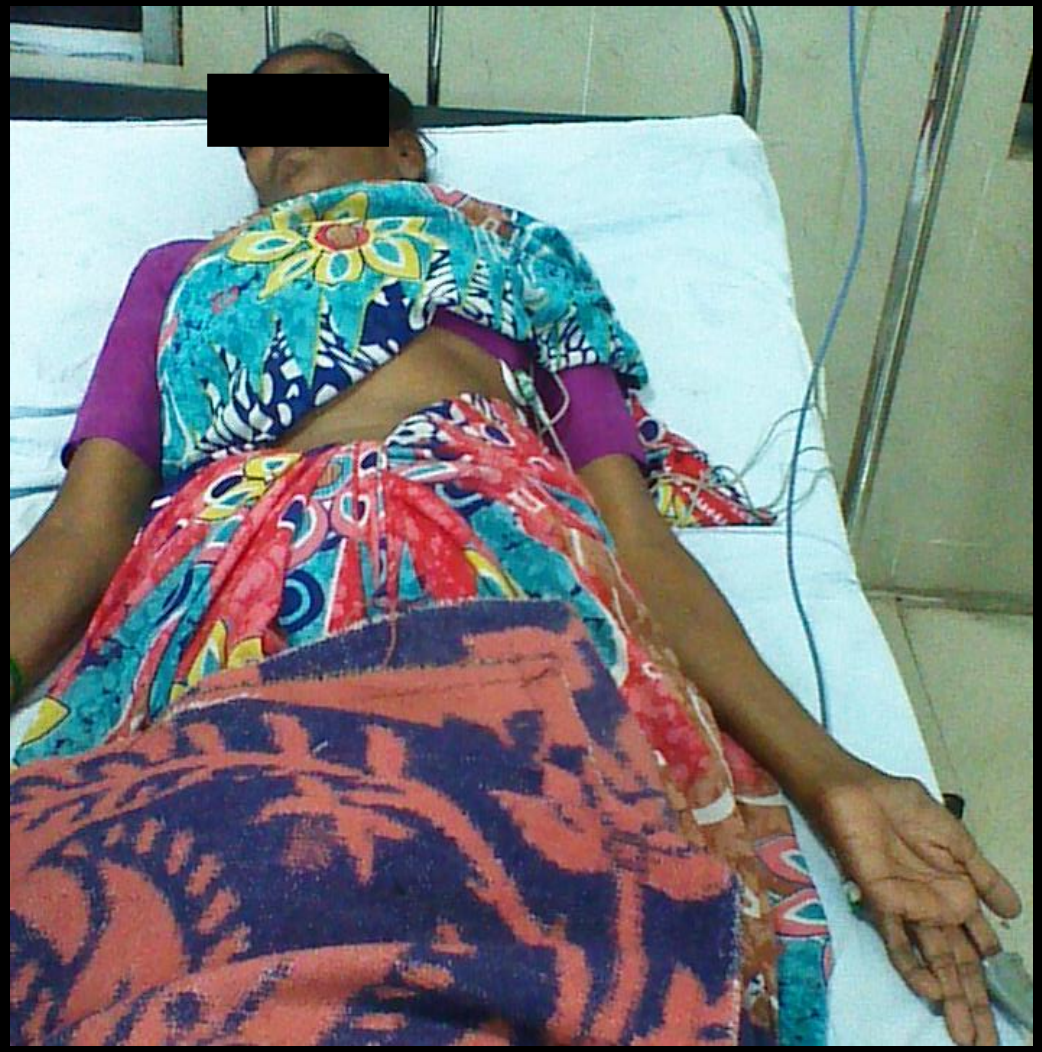
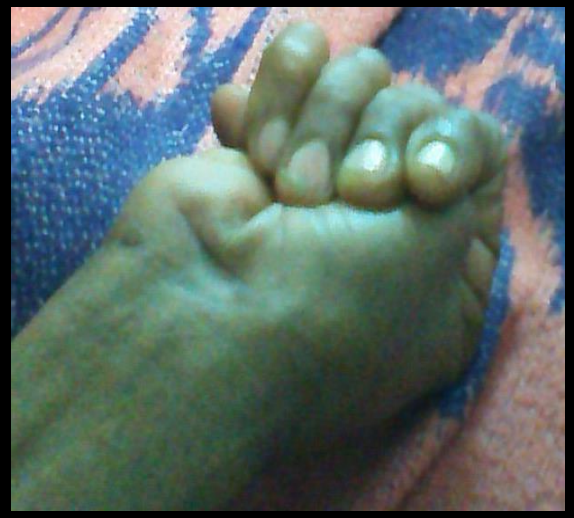
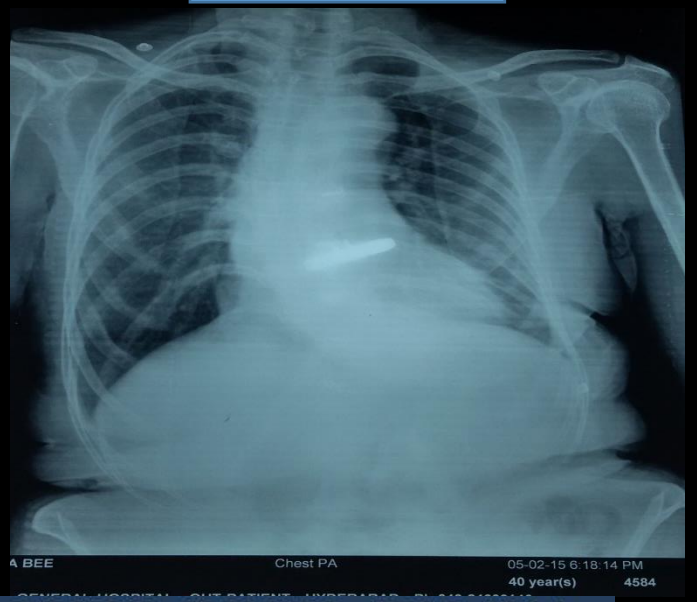
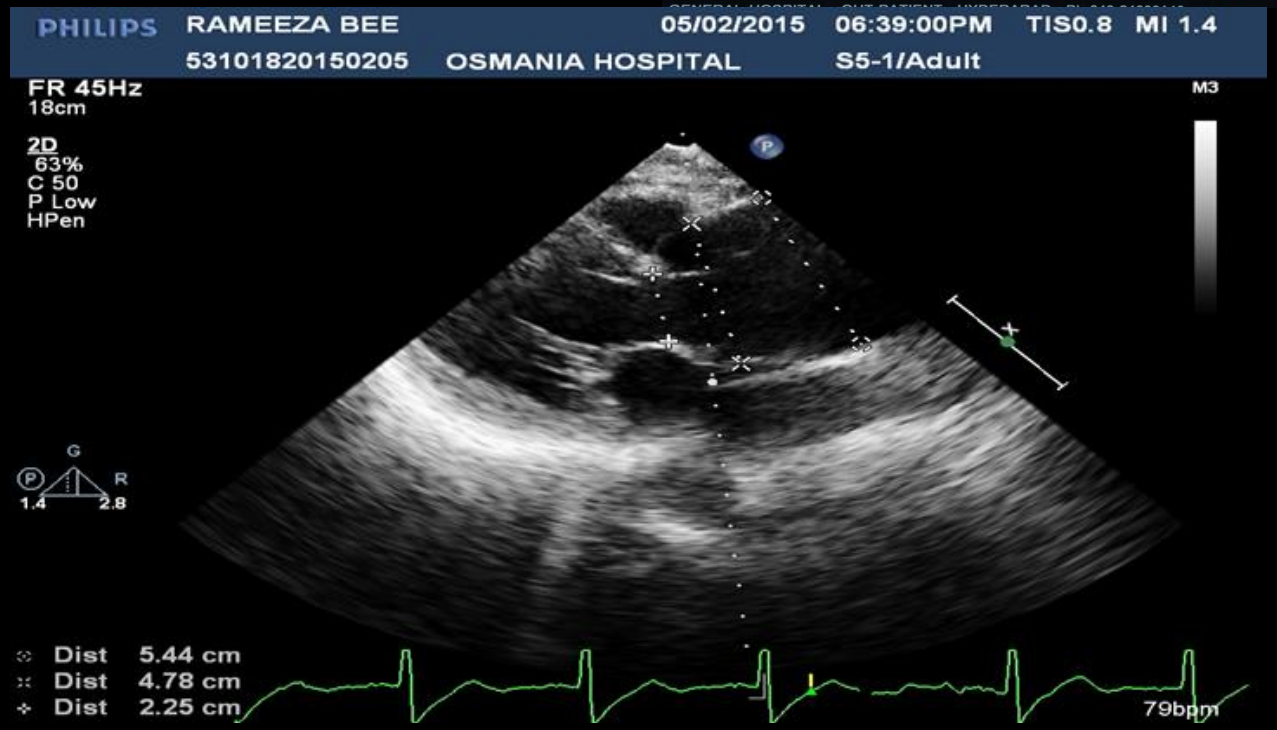


Figure .2

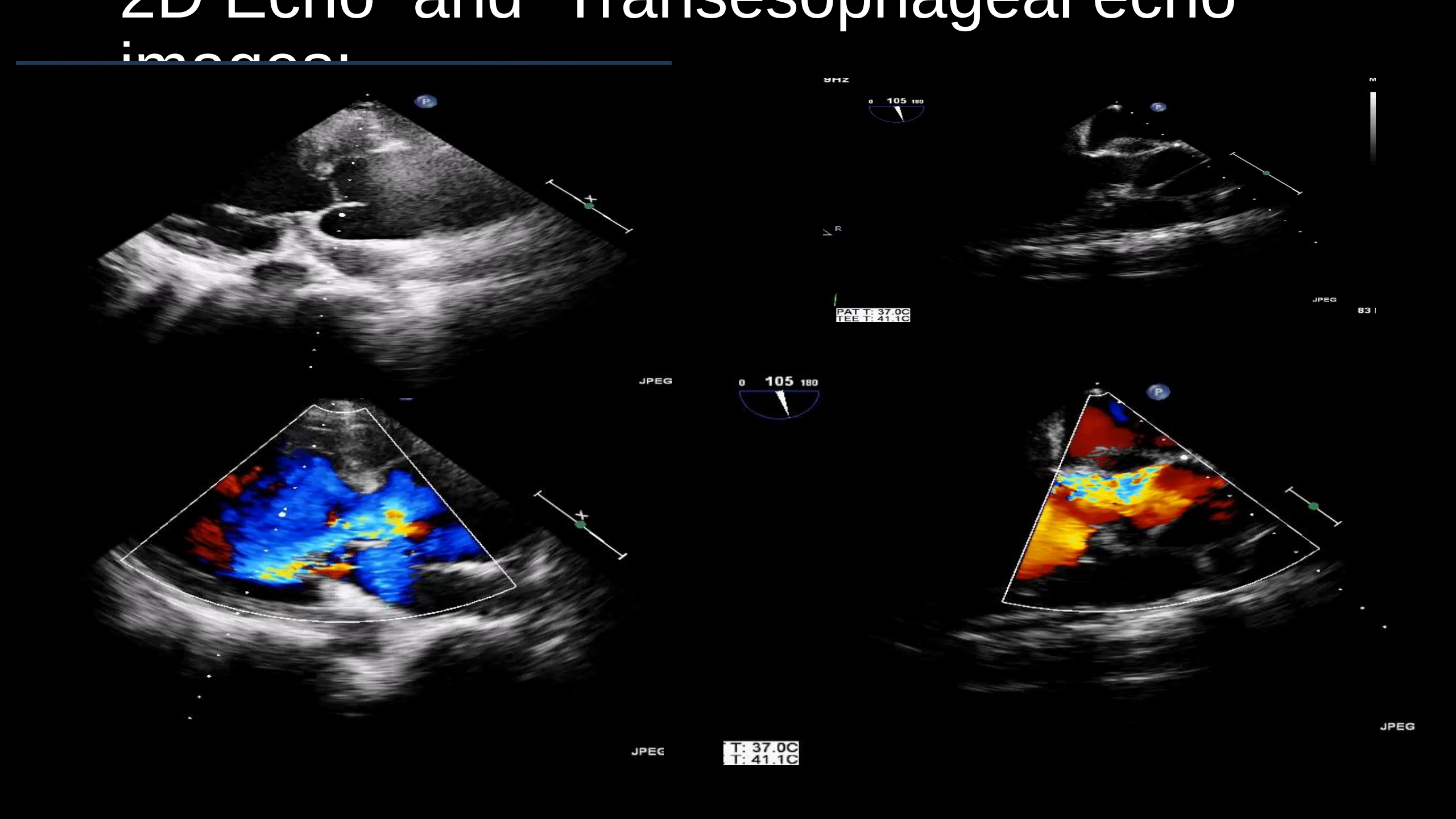


Chest x ray.3





2D Echo and Transesophageal echo images:



Diagnostic Criteria for Marfan syndrome:

	Cardiovascular	Skeletal system	Ocular system	Skin and integument	Pulmonary system	Family history/ genetic	dura
Major criteria	Dilation of ascending aorta (+AI),involving sinus of Valsalva. AAD	Pectus carinatum, Pectus excavatum, Reduced upper to lower segment ratio , increased arm span to height ratio, positive wrist and thumb sign, elbow extension reduced < 170 ,pes planus , protusion acetabulae.	Ectopia lentis	none	none	Parent ,sibling,child with Marfan disease,FBN1 mutation	Dural ectasia
Minor criteria	MVP, dilation of main pulmonary artery < 40yr of age.	Pectus excavatum ,hypermobile joints,crowding of teeth, high arched palate	Flat cornea, increased axial length of globe	Striae Recurrent or spontaneous hernia	Spontaneous pneumothorax, Apical bullae	none	none

For index case no mutation:
Major criteria in two organ system+
Involvement of another organ system

For index case –documented mutation:
Major criteria one organ system +
Involvement of another organ system.

Relative known case:
Major criteria in the family history +
Major criteria one organ system +
Involvement of another organ system

An international panel of experts revised the criteria in 2010 :

- New diagnostic criteria puts more weight on the cardiovascular manifestations of the disorder. Aortic root aneurysm and ectopia lentis (dislocated lenses) are now cardinal features:
- Aortic root dilatation (Z-score ≥ 2 when standardized to age and body size)

In the absence of family history:

1. **Aortic Root Dilatation Z score ≥ 2 AND Ectopia Lentis = Marfan syndrome.**
2. **Aortic Root Dilatation Z score ≥ 2 AND FBN1 = Marfan syndrome**
3. **Aortic Root Dilatation Z score ≥ 2 AND Systemic Score ≥ 7pts = Marfan syndrome.**
4. **Ectopia lentis AND FBN1 with known Aortic Root Dilatation = Marfan syndrome.**

In the presence of family history:

1.Ectopia lentis AND Family History of Marfan syndrome = Marfan syndrome .

2.A systemic score ≥ 7 points AND Family History of Marfan syndrome = Marfan syndrome.

3. Aortic Root Dilatation Z score ≥ 2 above 20 yrs. old, ≥ 3 below 20 yrs. old) + Family History of Marfan syndrome = Marfan syndrome

Discussion:

- The incidence of aortic dissection is estimated to be 2-3.5/10,000 persons per year and peak incidence at sixth and seventh decade with overall mortality 1% /hour .
- Patients with marfans syndrome are at higher risk for aneurysm of aorta , dissection can occur at younger age.It is classified into de Bakey type 1,2,3 and Stanfords type A and B depending upon location of dissection.
- High clinical suspicion required for diagnosing aortic dissection .It has variable clinical manifestations most common is chest pain (80%),severe aortic insufficiency (45%),hypotension(14%),shock (13%),syncope (12%),MI(7-19%),CVA(8%) and paraplegia (2%),pulse deficit (26%).
- TTE sensitivity 77-80% , specificity 93-96% and TEE has sensitivity 98% , specificity 95% respectively.
- One major limitation – artifacts that mimic as flaps(mirror image artifact)
- In our patient, chest pain was present along with aortic insufficiency with no pulse deficit.
- The management of the dissection: beta blockers are the drug of choice, followed by ACE inhibitors, Medical management is considered in uncomplicated and chronic type B dissection.
- Surgery is the treatment of choice for acute type A, complicated type B, associated with Marfans syndrome, end organ dysfunction.
- Endovascular therapy can be done as an alternative in complicated type B dissections.

Final Diagnosis:

- Marfan Syndrome
- Ascending Aorta and arch aneurysm
- Stanford Type A aortic dissection, Severe Aortic Regurgitation.
- Sinus Rhythm
- Good LV Function
- No CCF

Conclusion:

- Marfan syndrome – rare disease variable presentation with major cardiovascular manifestation.
- Aortic aneurysm and dissections incidence more common than general population .
- Early clinical recognition and prompt management of aortic dissection in these patients will have favorable outcome.

Reference:

1.Hurt's the Heart,13th edition: Mc Gram Hill; page 2261-2282.

2. J med genet 2010 ; 47: 476-485 – the revised Ghent nosology for Marfan syndrome.

3. Hiratzka et. al Guidelines for the Diagnosis and Management of patients with Thoracic Aortic Disease- Circulation.2010;121:e266-e369.