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 function.

- TEE: Aortic root aneurysm with dissection of aorta type-A with dissecting flaps

- Referred to CT surgery department for BENTALL procedure.
2D Echo and Transesophageal echo images:
## Diagnostic Criteria for Marfan Syndrome:

<table>
<thead>
<tr>
<th>Major Criteria</th>
<th>Cardiovascular</th>
<th>Skeletal System</th>
<th>Ocular System</th>
<th>Skin and Integument</th>
<th>Pulmonary System</th>
<th>Family History/Genetic</th>
<th>dura</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dilation of ascending aorta (+AI), involving sinus of Valsalva. AAD</td>
<td>Pectus carinatum, Pectus excavatum, Reduced upper to lower segment ratio, increased arm span to height ratio, positive wrist and thumb sign, elbow extension reduced &lt; 170, pes planus, protusion acetabulae.</td>
<td>Ectopia lentis</td>
<td>none</td>
<td>none</td>
<td>Parent, sibling, child with Marfan disease, FBN1 mutation</td>
<td>Dural ectasia</td>
<td></td>
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<td>MVP, dilation of main pulmonary artery &lt; 40yr of age.</td>
<td>Pectus excavatum, hypermobile joints, crowding of teeth, high arched palate</td>
<td>Flat cornea, increased axial length of globe</td>
<td>Striae</td>
<td>Spontaneous pneumothorax, Apical bullae</td>
<td>none</td>
<td>none</td>
<td></td>
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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 ≥ 7 pts = 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:


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