Case Study on Marfan's Syndrome and Bentall Procedure

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Introduction

Marfan syndrome is an inherited disorder that affects connective tissue — the fibers that support and anchor the organs and other structures in the body. Marfan syndrome most commonly affects the heart, eyes, blood vessels and skeleton. The damage caused by Marfan syndrome can be mild or severe. If the heart or blood vessels are affected, the condition can become life-threatening.[1]

Case discussion

Mr. X, 44 years old Male, Farmer got admitted in the Cardiac Care Unit(CCU) with the complaints of Breathlessness for past 1 year, and it is increased for the past one month, NYHA (New York Heart Association) dyspnea grade II moved to grade III in past 6 months and extra diastolic murmur was present. After History Collection, Physical Examination and Echo he was diagnosed as a case of Marfan's syndrome, Aortic Root Dilation with Moderate AR (Aortic Regurgitation) and Large Bullae right lung lower and upper lobe.

Past Medical History

Mr. X had lung Tuberculosis 10 years back and was treated with ATT (Anti Tuberculosis Drugs) for 12 months. He is not a known case of diabetes mellitus and hypertension.

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Family History

There is no family history of any specific heart disease and other communicable and non communicable diseases.

Personal History

Mr. X had the history of smoking and alcoholism since 15 years old and he stopped both smoking and alcohol at the age of 35. No history of allergic to any food and drugs.

Surgical History

Mr.X has the past history of hydrocele eversion and hemorrhoidectomy. Presently Mr.X has undergone right posterior lateral thoracotomy, right lower and upper lobe bullectomy also had Right side ICD(Inter Costal Drainage) and underwent Bentall's procedure (replacement of the aortic valve, root and the entire ascending aorta) for the aortic dilation.

Investigations on admission

Weight: 59 Kgs

Height : 180 Cms

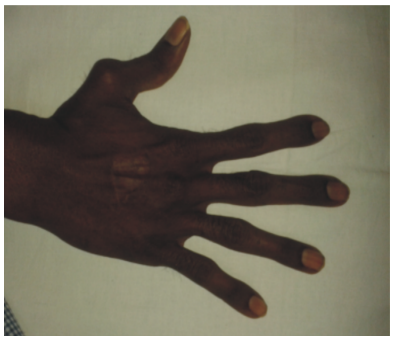
BMI: 18.2 Kg/ m^2

Arm length: 192 Cms

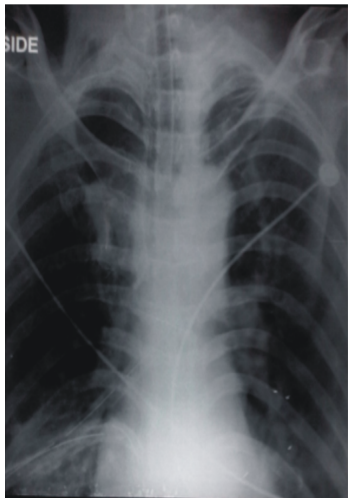
Physical examination:

Echo cardiogram : Aortic Aneurysm – proximal ascending aorta and aortic arch, no dissection flap, moderate aortic regurgitation, ejection fraction – 60%, normal left ventricular systolic function, ascending aorta 45 mm, aortic arch 44 mm dilated.

Thin fingers



Chest x ray: Large Bullae right lung lower and upper lobe and dilated aorta, patient has chest drainage right side lung



Marfan's Syndrome

Definition: Marfan syndrome is a disorder of connective tissue, the tissue that strengthens the body's structures. Disorders of connective tissue affect the skeletal system, cardiovascular system, eyes, and skin.[1]

Bentall Prosthesis

If the aorta enlarges to a certain size (about 2 inches [5 centimeters]), it is usually treated surgically. A Bentall procedure is a cardiac surgery operation involving composite graft replacement of the aortic valve, aortic root and ascending aorta, with re-implantation of the coronary arteries into the graft. This operation is used to treat combined aortic valve and ascending aorta disease, including lesions associated with Marfan syndrome.[7]

Post operative advice

Post operative annual assessment of entire aorta by MRI.[2]

Conclusion

Spontaneous new gene mutations leading to Marfan (less than 1/3 of cases) cannot be prevented. Patients with Marfan's syndrome should consult their doctor at least once every year. Heart related complications may shorten the lifespan of people with this disease. However, many people live into their 60s. Good care and surgery may further extend lifespan.[8]

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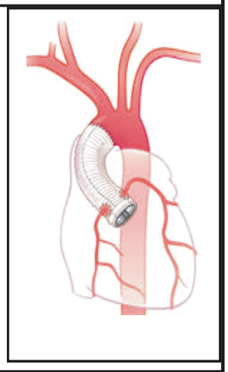
Book picture	*Patient picture*
Causes: • Defects in a gene called fibrillin-1 • Inherited • Up to 30% of patients have no family history (8,9)	• Patient has no family history
Incidence: 5 in 1,00,000 population worldwide (2)	
Signs and symptoms: • Usually tall with long, thin arms and legs and fingers. • When they stretch out their arms, the length of their arms is greater than their height. • Funnel chest (pectus excavatum) or Pigeon breast (pectus carinatum) • Flat feet • Highly arched palate and crowded teeth • Hypotonia • Joints that are too flexible (but the elbows may be less flexible) • Learning disability • Movement of the lens of the eye from its normal position (dislocation) • Nearsightedness • Small lower jaw (micrognathia) • Spine that curves to one side (scoliosis) • Thin, narrow face • Heart murmurs (4,6)	• Patient is tall and has thin arms and legs and fingers, BMI: 18.2 Kg/ m^{2}- underweight, • Height : 180 Cms, Arm length: 192 Cms • Pigeon breast • Flat feet • Highly arched palate and crowded teeth • Small lower jaw (micrognathia) • Thin, narrow face • Extra diastolic murmur present
Medical Management: • Beta blockers • Losartan - blood pressure lowering drugs to help prevent the aorta from enlarging and to reduce the risk of dissection and rupture (4) **Surgical management:** • Aortic repair: Many physicians have adapted the criterion of a 50 mm maximum aortic root dimension for performing elective surgery in adult patients with Marfan's syndrome.(5, 10)	Losartan 25 mg OD Aortic repair – **Bentall procedure** (Patient had 45 mm dilated aortic arch) 
Complications • Aortic regurgitation • Aortic rupture • Bacterial endocarditis • Dissecting aortic aneurysm • Enlargement of the base of the aorta • Heart failure • Mitral valve prolapse • Scoliosis • Vision problems (3)	• Moderate aortic regurgitation • Aortic Aneurysm – proximal ascending aorta and aortic arch - Ascending aorta 45 mm, aortic arch 44 mm dilated

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