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Case Note

Marfan's syndrome with aortic valve endocarditis

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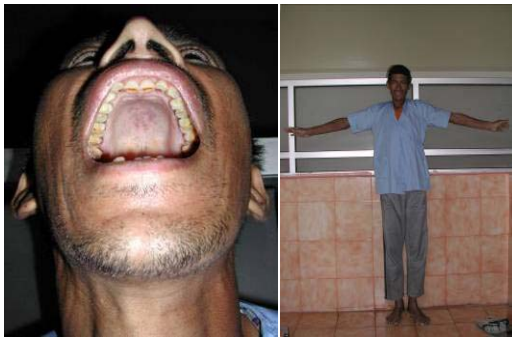
Abstract
Marfan's syndrome is an Autosomal dominant disorder of the connective tissues resulting in abnormalities of the musculoskeletal system, cardiovascular system and eyes. It has a prevalence of 1 in 100,000 population[1] and occurs in all ethnic groups. It may be familial or due to new mutation (30%), in the fibrillin gene on arm of chromosome 15. It is estimated that one person in every 3000-5000 has Marfan's syndrome may have cardiovascular abnormalities and may be complicated by infective endocartditis. About 90% of Marfan patients will develop cardiac complications[2].
The patient under discussion has musculoskeletal (Tall stature, reduced upper-lower segment ratio, arm-span to height ratio >1.05, high arched palate) and Cardiovascular features (Severe aortic regurgitation complicated with infective endocarditis)

Key words: Marfan's Syndrome, Autosomal dominant, Aortic Regurgitation, Infective Endocarditis.

Marfan's Syndrome is an inherited disorder of the connective tissue that causes abnormalities of the patient's musculoskeletal system, cardiovascular system and eye. It is named for the French pediatrician, Antoine Marfan (1858-1942), who first described it in 1896. Marfan's Syndrome is sometimes called arachnodactyli, which means "Spider-like fingers" in Greek, since one of the characteristic signs of the disease is disproportionately long fingers and toe. It is estimated that one person in every 3000-5000 has Marfan syndrome, which can be complicated by cardiovascular abnormalities and further can be complicated by infective endocarditis.

Case Report
Mr. S.D 35 year old farmer from Saranthali, Hindu by religion was admitted in Medical ward of Kathmandu Medical College & Teaching Hospital on 2060/10/10 with the chief complain of high grade fever, continuous type associated with chills and rigor and generalized body ache of 11 days durations. He had no major medical or surgical history in the past.

General Examination revealed marfanoid features: Tall stature (184cm), reduced upper-lower segment ratio (86:98cm), arm-span>total height (197>184cm i.e. 1.07), high arched palate, petichae in the palate, arachnodactyli with positive thumb sign (Sternberg) and wrist sign (Murdoch), other positive findings were pedal oedema. His vitals showed temperature of 102^{o} F, pulse of 110/min regular and BP 100/40 mm of Hg.



Systemic examination revealed early diastolic murmur heard at left sternal edge with patient leaning forward breath held in expiration (3/6 grade) and on day 17 of admission murmur changed its characteristic where it became a musical murmur (Sea Gull Murmur).

Investigation
Total white cell counts: 25700/mm^3 ($P_{88\%}$, $L_{11\%}$), after a week of treatment the total white cell count was 15800/mm^3 ($P_{81\%}$, $L_{11\%}$), again after 3 weeks of treatment the total white cell count reduced to 8800/mm^3 ($P_{70\%}$, $L_{22\%}$). Hb%-8.5gm%, ESR-60, CRP-Positive.

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Blood Culture- 3 samples half an hour apart were taken from 3 different sites for bacteria and fungus showed no growth.

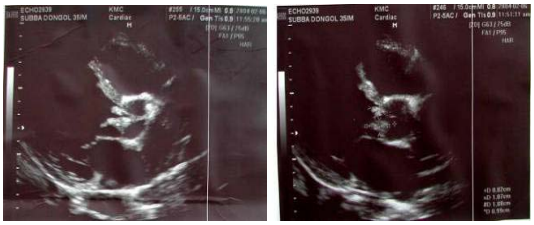
Random blood sugar, blood urea, serum creatinine were in normal range.

Urine routine examination: Albumin trace, RBC- Nil.

X-ray:- Cardiomegaly of left ventricular type.

ECG: - Sinus tachycardia (PR interval—0.20 Sec)

ECHO: - Moderate to severe aortic regurgitation, vegetation in RCC and NCC of 1.07cm x 0.82cm aortic valve, dilated left atrium (4.2cm) and left ventricle (5.9cm). Repeat ECHO on day 17th of admission showed: - Old findings, No perforations or abscess, but associated with mitral valvulitis and normal left ventricle function.



Considering all the clinical and laboratory findings, diagnosis of Marfan's syndrome with aortic regurgitation with infective endocarditis and CCF was made.

Treatment

Patient was managed with salt restriction, bed rest, Crystalline Penicillin 4 million IU 4 hourly through central line, Gentamicin 60mg TID, Diuretics, Digoxin and ACE Inhibitor (Enalapril).

After 2 weeks of treatment the fever didn't subside and Cefazoline 1gm QID was started but the fever still persisted so the patient was transferred to Shahid Gangalal National Heart Centre (SGNHC). There he was started on Vancomycin for 2 days but he couldn't afford it so was changed to Injection Cloxacillin 2gm QID, which he received for 42 days. Once his clinical status improved, he underwent aortic valve replacement (AVR) during his stay at SGNHC and had an uneventful recovery.

Discussion

Marfan syndrome is an inherited disorder of the connective tissue that causes abnormalities of the patient's eyes, cardiovascular system, and musculoskeletal system. It is named for the French pediatrician, Antoine Marfan (1858-1942), who first described it in 1896. Marfan syndrome is sometimes called arachnodactyli, ("spider-like fingers" in Greek) since one of the characteristic signs of the disease is disproportionately long fingers and toes. It is estimated that one person in every 3000-5000 has Marfan's syndrome. It may be accompanied by cardiovascular abnormalities and be further complicated by infective endocartditis.

Causes

Marfan syndrome is caused by a single gene for fibrillin on chromosome 15, which is inherited in most cases from an affected parent. Between 15 and 25% of cases result from spontaneous mutations. Mutations of the fibrillin gene (FBNI) are unique to each family affected by Marfan, which makes rapid genetic diagnosis impossible, given present technology.

Symptoms

Cardiac and circulatory abnormalities

- **Aortic enlargement**. This is the most serious potential complication of Marfan syndrome. Because of the abnormalities of the patient's fibrillin, the walls of the aorta (the large blood vessel that carries blood away from the heart) are weaker than normal and tend to stretch and bulge out of shape. This stretching increases the likelihood of an aortic dissection, which is a tear or separation between the layers of tissue that make up the aorta.
- **Aortic regurgitation.** A weakened and enlarged aorta may allow some blood to leak back into the heart during each heartbeat; this condition is called aortic regurgitation.
- **Mitral valve prolapsed** Between 75 and 85% of Marfan patients have loose or "floppy" mitral valves, which are the valves that separate the chambers of the heart. When these valves do not cover the opening between the chambers completely, the condition is called mitral valve prolapsed.
- **Infective endocarditis.** Infective endocarditis is an infection of the endothelium, the tissue that lines the heart. In patients with Marfan, it is the abnormal mitral valve that is most likely to become infected.

Musculoskeletal abnormalities

- Scoliosis.
- Kyphosis.
- Pectus excavatum.
- Pectus carinatum.
- Protrusio acetabulae.

Disorders of the eyes

- Ectopia lentis.
- Glaucoma.
- Cataracts.
- Retinal detachment.

Other disorders

- Striae.
- Obstructive sleep apnea.

Diagnosis

Diagnostic Criteria according to the GHENT NOSOLOGY.

The diagnosis of Marfan's syndrome with classical phenotype remains largely clinical. The diagnosis is made by taking a family history and through examination of the patient's eyes, heart, and bone structure. The examination should include an echocardiogram taken by cardiologist, a slit-lamp eye examination by an ophthalmologist, and a work-up of the patient's spinal column by orthopedic specialist.

New investigation is **mutation analysis** (study of FBNI gene).

Treatment

The treatment and management of Marfan is tailored to the specific symptoms of each patient. Some patients find that the syndrome has little impact on their overall lifestyle; others have found their lives centered on the disorder.

Cardiovascular system

After a person has been diagnosed with Marfan, he or she should be monitored with an echocardiogram every six months until it is clear that the aorta is not growing larger. After that, the patient should have an echocardiogram once a year.

Musculoskeletal system

Children diagnosed with Marfan should be checked for scoliosis by their pediatricians at each annual physical examination. Pectus excavatum and pectus carinatum can be treated by surgery.

Ocular system

Patients with Marfan should have a thorough eye examination, including a slit-lamp examination, to test for dislocation of the lens as well as nearsightedness.

Social and lifestyle issues

Smoking: Smoking is particularly harmful for Marfan patients because it increases their risk of emphysema.

Pregnancy: Until very recently, women with Marfan were advised not to become pregnant because of the risk of aortic enlargement or dissection. The development of beta-blockers and echocardiograms, however, allows doctors now to monitor patients throughout pregnancy. It is recommended that patients have an echocardiogram during each of the three trimesters of pregnancy. Normal, vaginal delivery is not necessarily more stressful than a Caesarian section, but patients in prolonged labor may be given a Caesarian to reduce strain on the heart. A pregnant woman with Marfan should also receive genetic counseling regarding the 50% risk of having a child with the syndrome

Prognosis

The prognosis for patients with Marfan has improved markedly in recent years. As of 1995, the life expectancy of people with the syndrome has increased to 72 years, up from 48 years in 1972. This dramatic improvement is attributed to new surgical techniques, improved diagnosis, and new techniques of medical treatment.

The most important single factor in improving the patient's prognosis is early diagnosis. The earlier that a patient can benefit from the new techniques and lifestyle modifications, the more likely he or she is to have a longer life expectancy.

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