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RESEARCH ARTICLE

CASE STUDY ON MARFAN SYNDROME

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Abstract

Introduction:-

Mar fan syndrome is an autosomal dominant, multisystem connective disease, associated with mutation in fibrillation. It mostly affects skeleton, lungs, eyes, heart, and the aorta Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectus excavatum, pectus carni vatum, or ectopia lens in eyes. The incidence of mitral prolapse in such patients is essentially equal in children and adults of the same sex.

A patient with marfanoid habitus was admitted to the Sree Balaji Medical College and Hospital, Chennai in April 2015 for surgical closure of an atrial septal defect. He was suspected to have Marfan syndrome but there was no involvement of the aorta nor the eye. The clinical features were intermediate between that of the Marfan Syndrome It is suggested that this could be a separate distinct entity within the heritable disorders of connective tissue known as the Marfanoid Syndrome.

Objectives:-

The objective of this study is to emphasize the difficulties encountered in making a diagnosis of the Marfan syndrome and to review some of the diagnostic aids in individuals suspected of having this syndrome where only equivocal features are .present.

Background:-

Worldwide, the incidence of Mar fan syndrome is approximately 7–17/100,000 .The incidence of aortic dilatation and mitral prolapse in patients with Marfan's syndrome was essentially equal in children and adults of the same sex. The oral cavity shows high-arched palate that result from a narrow maxilla .Connective tissue disorders have been associated with severe periodontitis. Here I am presenting a case report of Marfan syndrome with dental decay which is successfully treated.

Case report:-

The patient, a 26-year-old arumugam young man, was first seen in sree balaji medical college and hospital on 6th April 2015.after being referred from SMC for consideration of prosthetic aortic valve implantation. As far as could b ascertained, his development, except that he was taller than the rest of his family members and friends, has been

entirely normal, until the age of 18, when he was first diagnosed to have Marfan syndrome. No family history of such disease was present; everyone else seems to be fine according to the patient with no significant medical conditions, except for the mother, who suffers from G6PD. Since the age of 6, the patient suffered from severe myopia and underwent surgical correction accordingly. In 1995, surgical lens. Removal for the management of congenital myopia and IOL implant were done on the patients left eye at vasan eye care Hospital, Chennai.

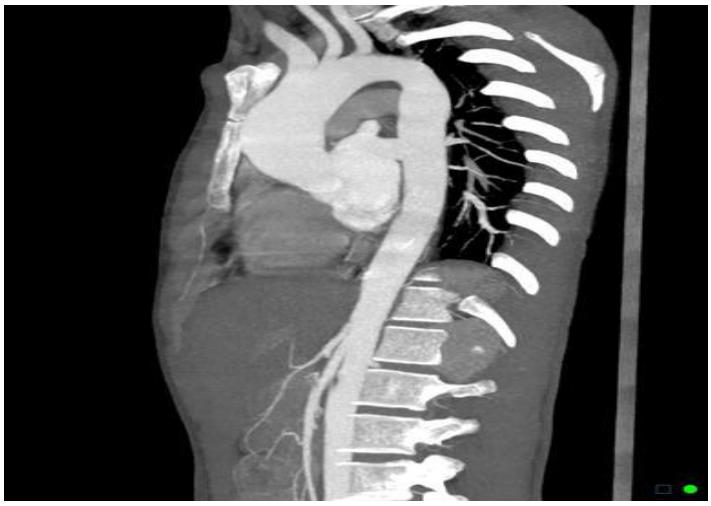
Another surgery (Sclera buckle surgery) was done on the same eye in 2011 for the management of retinal detachment. The patient also underwent a similar surgery at the age of 6 for his right eye, but the surgery wasn't successful, and as a result the patient suffered from right-eye blindness afterwards. On April 27th, the patient noted a mild productive but gradually progressive cough, with a high grade fever (around 40 degrees), a mild shortness of breath, fatigue and night sweats. The patient complained of severe dyspnea and chest pain when lying flat on his back or laterally on his left side. However, no headache, syncope, nausea or vomiting was noted.

The patient went for checkups the following day, and was prescribed antibiotics. Fever was still high, and not responsive to the antibiotics, therefore the patient paid another visit to the doctor, and proper investigations showed a severe chest infection. The patient was given pain relief medications, cough medications and antibiotics and then he returned back to sree Balaji Medical College and hospital on May 4th, where he was admitted in INTENSIVE CARE UNIT for his chest infection. On routine evaluation he was found to have a dilated mediastinum, and on further evaluation, he was found to have an aortic dissection. Hence he was referred to VASAN EYE CARE for further investigations. The patient is unmarried, unemployed, and lives with his parents. He is a never smoker and a never drinker, with no known allergies.

Physical examination revealed him to be a tall, thin white man with extremely long fingers, who appeared much older than his stated age. Blood pressure was 160/100 mmHg, the pulse was 110 beats per minute, with a regular rhythm and a collapsing character. The patient had a temperature of 37.8 and was slightly tachypneic with a respiratory rate if 22 breaths per minute. Closer examination of the hands showed stage 3 clubbing (increased curvature of the nail bed), some arthritic changes such as the Z-thumb deformity. Characteristic Marfan bony changes were also present, in which wrist and thumb signs were noted as well as a mild pectus excavatum. The patient suffered from a high arched palate but no dental abnormalities were present. Upon assessment of the precordium, a hyperdymanic displaced apex beat was felt in the 6th intercostal space, midclavicular line. A grade 4 early diastolic murmurs was heard over the tricuspid and aortic areas, but it was with a greater intensity over the tricuspid region. S1 and S2 were both audible, however S2 was louder. On percussion and auscultation of the back, right basal crackles were heard with dullness over the right lower lobe of the lung. Examination of the abdomen showed visible pulsation above the umbilicus, and stretch marks. There was no venous distension, organomegaly, cyanosis, or ascites. Peripheral pulses were felt, and no edema was detected in the lower limbs. On systematic review, no abnormalities were found other than the patient's presenting symptoms, with episodic headaches and dizziness, and some mild joint pain at the knees. Regular tests were first carried out, such as CBC, U&E, and LFTs.

X-rays were done and showed a right basal consolidation with cardiomegaly .CT was also done and showed a very large aneurysm affecting the ascending thoracic aorta starting at the level of the aortic valve and terminating just proximal to the arch, measuring approximately 9x7cm in maximum dimension. The dissection was shown to be starting very low at the level of the valve and involving the entire aneurysmal aorta, preserving the arch. There was no involvement of the arch or the major vessels arising from the arch. The remainder of the thoracic aorta including the arch, the descending thoracic aorta and also the abdominal aorta were all normal in caliber with no evidence of dissection. The iliac arteries were within normal limits. There was a consolidation noted affecting the right lower lobe and also some pleural fluid was noted in the right side. Some fluid was also detected in the right hilum which was felt to be reactive. No CT evidence of aortic rupture was shown. A gross cardiomegaly was also visible.

Echo was done twice, one before the surgery and one after, to allow for comparison and to assess the patient's improvement. The pre-op echo showed a severe aortic regurgitation, a mild mitral prolapse with a minor mitral regurgitation and a trivial pulmonary regurgitation. A severe dilatation of the aorta (6.8 cm) and sinus of valsalva (6.6 cm) with a type (A) dissection flap of the ascending aorta were seen. The post-op (Bentall procedure) echo showed a significant difference. Prosthetic valve was well seated with normal motion, and no significant aortic incompetence or mitral incompetence.



After initially controlling the patient's chest infection, the patient underwent Bentall surgery on May 10th for dissection of ascending aorta , a severe aortic regurgitation, and an aortic aneurysm of around 8 cm. The surgery was performed through mediansternotomy and vertical pericardiotomy. After cardioplegia, aortotomy was done with resection of the aneurysm and the AV. Coronary arteries were dissected with 10.0mm buttons. Replacement of valve was done using 29 carboseal valsalva bileaflet composite grafts. This is the worldwide used procedure for such indications.

During the post-op phase, 10 packs of cryoprecipitate, and 2.4 mg IV of factor 7 were given to maintain the patient hemo dynamically. The patient was also given morphine and IV panadol to relieve the pain. A mild productive but gradually improving cough with white sputum was present. The patient was managed with medication throughout his hospital stay.Warfarin dosage was given depending on the INR.

Discussion:-

Pathophysiology and etiology:-

Fibrillin is an important component of the microfibrillar system that acts as a scaffold for elastogenisis. Classical Marfan syndrome is associated with a mutation in FBN1, the gene that encodes for fibrillin-1. The pathophysiological outcomes of the degeneration of elastic fibers in Marfan syndrome seem to explain the majority of manifestations of this condition. Stiffness and reduced distensibility of the aorta in response to increased pulse pressure, is the main most important consequence of elastin degeneration.5 recently, another hypothesis has emerged trying to explain the pathophysiology behind Marfan syndrome. Transforming growth factor β (TGFβ), a cytokine that regulates cell morphogenesis, is thought to contribute to the Marfan syndrome phenotype. Abnormal fibrillin causes failure of the sequestration of the inactive latent precursor of TGFβ, resulting in excessive TGFβ activation, and thus producing the phonotypical manifestations of Marfan's.

Clinical presentation:-

Marfan syndrome primarily involves the skeletal, ocular and cardiovascular systems. Typically patients with Marfan syndrome present with tall stature, ectopia lentis, aortic root dilatation, and positive family history. Our patient presented with all the mentioned symptoms except for the family history.

Differential diagnosis:-

Clinical diagnosis of Marfan syndrome is challenging because of the increased marfenoid features of other connective tissue diseases. Differential diagnosis could include homocystinuria, familial aortic dissection, familial arachnodactaly, Ehler Danlos syndrome and MEN IIb. Serum methionine must be carried out to rule out homocystinuria.3 Molecular techniques have not been undertaken widely as a method to distinguish between Marfan syndrome and other similar-featured disorders, as it is not clear whether they can differentiate between those conditions with overlapping symptoms.5

Management:-

Although clinical management of genetic disorders is not backed up by extensive clinical trials on humans, numerous studies conducted in vivo managed to establish a direct link between the administration of angiotensin

receptor blockers (ARBs) and the inhibition of TFGβ signaling.4 Various retrospective studies have assessed the beneficial effects of beta blockers (BB) therapy in Marfan syndrome, and considered it to be the standard of care. The potential benefit of beta blockers isattributed to the reduction of aortic wall stress and heart rate.8 All Marfan syndrome patients who can tolerate beta blockers should be treated regardless of the presence or absence of aortic dilatation. No randomized trials reported solid evidence on the use of angiotensin converting enzyme inhibitors (ACEI), however this class of drugs has the theoretical advantage of reducing the ejection impulse and vascular smooth muscle apoptosis which is implicated in cystic medial degeneration.5 Recent trials have been aiming to manufacture drugs that are directed at the fibrillin-1 or TGFβ axis to produce the maximum desirable effect.

Solution:-

Solutions such as contact lenses or pectus repair. Finally, it may be useful to identify a role model or age-matched peer with the Marfan syndrome to discuss frustrations and opportunities with an affected child

The eye evaluations should be performed every year. It is essential to identify and correct high refractive error or amblyopia in childhood in order to preserve and maximize visual function. Individuals with the Marfan syndrome are at increased risk for glaucoma, cataract formation, and retinal detachment, even in the absence of ectopia lentis. Progression of skeletal abnormalities, especially scoliosis and anterior chest irregularity, can be dramatic during periods of rapid growth, such as puberty. Evaluation and follow-up by an orthopedist is indicated in these cases.

Recommendation:-

Many comparative studies have shown that there is a better outcome with early aortic root surgery than with an emergency or later surgery. Prophylactic surgery is recommended when the diameter at the sinus of Valsalva EXCEEDS 5.5 CM in adults.

Conclusion:-

Marfan syndrome is the most common inherited connective tissue disorder with diverse clinical manifestations. Although many studies have been conducted which aimed at improving the medical aspect of management, those trials produced conflicting results and generally involved relatively few patients.

This report underscores the importance of detailed family history and physical examination in the diagnosis of Marfan syndrome. Additionally, good insight about the pathogenesis and the clinical presentation of Marfan syndrome improve the effectiveness of medical therapies. Regular valvular monitoring and early initiation of beta blockers therapy as well as elective prophylactic surgical repair contribute to increasing the survival rate of Marfan patients.

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