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A Case Report on Marfan Syndrome

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ABSTRACT

Marfan syndrome is an autosomal dominant disorder of a connective tissue that mostly caused by mutation in the FBN1, the gene encoding fibrinillin1. The people affected by the Marfan syndrome are tall, slender and it also affects the organs like skeleton, lungs, heart, eyes, and the large blood vessel. A case Report of Marfan syndrome has been reported with chronic obstructive airway disease which was treated and resolved.

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INTRODUCTION

Marfan syndrome is also known as arachnodactyly. Marfan syndrome is an autosomal dominant disorder of a connective tissue that mostly caused by mutation in the FBN1, the gene encoding fibrinillin1. Marfan syndrome is a rare pleiotropic disease characterised by 3 clinical criteria- thoracic, aortic, aneurysm and/or dissection; ectopia lentis and multisystemic manifestations. And the disease is also chacterised by 2 genetic criteria [1]. The Marfan syndrome affects all over the body. But, mostly it effects on skeleton, eyes, heart and the aorta. The people affected by Marfan syndrome are tall and slender. They are having arachnodactyly scoliosis and a pectus excavatum, pectus carniatum or ectopial lentio in eyes. Several cases are appeared due to sporadic mutation in a single germ cell of the parent [2].

Causes

It was generally caused due to randomly occur faulty gene in the sperm or egg. The gene that carries Marfan syndrome is FBN1. The FBN1 provides a protein called fibrinillin-1. this fibrinillin-1 provides strength and elasticity to the connective tissue. Most of the times the Marfan syndrome was inherited from the parents to children [3].

Signs and Symptoms

The signs of Marfan syndrome include abnormally long, slender or spidery fingers and toes. The common symptoms of Marfan syndrome are myopia, topia lentis, retinal detachment, glaucoma, cataract formation, skeletal and connective tissue problems, joint laxity, dolichostenomalia, pectus exacatum, scoliosis, cardiovascular malformation, aortic root dilation, mitral wall prolapsed, and atrial septal defect, patent ductus areteriosus, pulmonary artery stenosis, and persistent left superior vena cava [4].

Epidemiology Marfan syndrome was first observed in 5.5 year old girl in 1896 by a French pediatrician named Antonin Marfan. The minimal birth incidence is around 1 in 9800 i.e. the progressive aortic dilation, usually maximal at the sinus of Valsalva, associated with aortic valve incompetence leads to aortic dissection or rupture and is the principal cause of mortality. But mitral valve prolapsed with in competence may be significant, and lens dislocation, myopia and arthritis associated with chronic joint laxity can cause substantial morbidity [5].

Pathophysiology

Marfan syndrome is associated with mutations in fibrillin-1, which is an important component of the elastic microfibril. The fibrillin-1 which is a 350kd glycoprotein normally synthesized as a 375kd precursor that is secreted into extracellular matrix. In extracellular matrix, it polymerizes to form microfibrils and helps to stabilize latent transforming growth factor-beta-binding proteins. This latent transforming growth –beta- binding proteins holds transforming growth factor beta in active state. If a failure occurs in the interaction between fibrillin-1 and LTBPs it may result in excess TGFβ signaling. Abnormal fibrillin or reduced amount of fibrillin may cause other problems of marfan phenotype. If different mutations are occurred in the same codon it may leads to severe neonatal Marfan syndrome. The mutations in the central area of the gene may be associated with severe phenotypes that ranging from neonatal Marfan syndrome to ectopia lentis. Increased TGFβ signaling was found in most patients with Marfan syndrome and TGFβ blockade by angiotensin-ii rescues the model phenotypes [6].

Diagnosis

Marfan syndrome was usually diagnosed by Ghent nosology. Generally in a young patient tall, thin body habits, long limbs and pectus deformities was observed. Along with these, sometimes arched palate with dental deformities, recurrent hernia, and pneumothorax was also observed [6].

Treatment

Management involves beta blockers to decrease the blood pressure and to prevent progressive dilation of the aorta. Angiotensin-II receptor blockers help to slow down the aortic root dilation. A non surgical measure involves glasses or contact lenses for near-sightedness. Back brace for people with scoliosis. Surgery may also be necessary to replace a faulty heart valve or to repair eye abnormalities, retinal detachments or cataracts [7].

CASE REPORT

A sixty year old male reported to the department of general medicine with chief complaints of shortness of breath since 4 days, frequent sore throat and cough. His medical history revealed that he has suffered from osteoarthritis and COPD which was resolved. The family

history of the patient was noncontributory. His social history revealed that he was smoker and alcoholic since 30 years. He is having poor oral hygiene along with that he is also having a cardiac disease.

The patient appeared tall stature with an average weight for his age and gender. He had disproportionately long arms and legs as compared with the track. Examination of his hands showed elongated fingers with thickened phalange joints. On examination of the foot it was revealed that he had flat feet with mild pronation along with elongated toes. Another finding was a subtly intended chest and mild hunching at back.

Investigations

The patient's lab data revealed abnormal hemoglobin levels i.e. 10gm/dl. His peripheral capillary oxygen saturation level (SPo2) was 90%. The ECG report shows inverted T-wave.

Differential Diagnosis

His long arms and legs, elongated fingers with thickened phalange joints represent Marfan syndrome. COPD was diagnosed by chest x-ray and arterial blood gas levels. The inverted T-wave may represent the conditions like: cardiac ischemia, pulmonary embolism.

Treatment

On the day of admission the patient was treated with inj.deriphylline-2ml (300mg)-IV- twice daily, inj.hydrocortisone-100mg-IV-twice daily, Neb. Duolin+ Budecort combination was given in SOS (When Ever Necessary). Along with this medication he was advised with Tab. Ranitidine-150mg –IV –twice daily. The treatment plan was continued for 10 days. On the very next day, he was prescribed with Tab. Escitalopram (10mg) + Clonazepam (0.5mg) – once daily as he had a complaint of insomnia.

DISCUSSION

Marfan syndrome is one of the most common lethal disease and was inherited in Mendelian fashion. It is a condition which affects the quality of life and can lead the person to frustration and low self-esteem. High nasal airway resistance also increases the susceptibility to obstructive sleep apnea. The cardiac pathology in these patients increases the risk of

endocarditis. As there is no particular therapy to treat Marfan syndrome the only way is to manage the underlying symptoms [8].

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