MARFANS SYNDROME-A CASE REPORT

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ABSTRACT

Marfan’s syndrome is a connective tissue disease inherited in an autosomal dominant manner and caused mainly by mutations in the gene FBN1, with an estimated prevalence of one in 10,000 to 20,000 individuals. This rare hereditary connective tissue disorder affects many parts of the body. The diagnosis of Marfan’s syndrome is established in accordance with a review of the following diagnostic criteria, known as the Ghent nosology, through a comprehensive assessment largely based on a combination of major and minor clinical manifestations in various organ systems and the family history. Aortic root dilation and mitral valve prolapse are the main presentations among the cardiovascular malformations of Marfan’s syndrome. Here with we are discussing a case of a 27 years old man who presented with typical features of Marfans syndrome.

KEYWORDS: Connective Tissue Disease, Marfans Syndrome, Aortic Root Dilatation

INTRODUCTION

This condition was first described in 1896, the French pediatrician antoine Bernard-Jean Marfan. It involves the multi organ system like cardiovascular, skeletal and Ocular systems, the integument, lungs and Dura. Marfan syndrome (MFS) is an autosomal dominant connective tissue disorder. A family history of Marfan’s syndrome has been found to be present in 49% of the families of individuals with this condition. In about 25-30% of the patients, the disorder occurs without a positive family history, and gene mutation is likely to be taken into consideration. Most such severe cases appear to be due to sporadic mutation in a single germ cell of one parent. Many familial cases may have milder manifestations (for instance, mitral valve regurgitation is less frequent) and has better prognosis, but may be more difficult to detect during infancy. The aortic root and arch diameters have been found to be significantly greater in patients with a family history than in those without such histories, and life expectancy has been found to be shorter. The patient also presents with typical facial and oral features as in the following case.

CASE REPORT

A male patient of 27 yrs old came to our o.p with a chief complaint of painful decayed tooth in upper back tooth region for the past 4 days. Pain is moderate, continuous and dull. On general examination Patient was tall in stature, head appeared to be dolicocephalic. Arachnodactyly was assessed using,walker Murdoch wrist sign. On extra oral examination drooping of eyelids and retrognathic mandible was seen. On intra oral examination high arched palate, crowding of teeth, supernumerary teeth was found. The patient was subjected to radiological investigation, opg revealed infected root stump in 17, periapical abscess in 13. On lateral cephalometric view malocclusion was seen.
was seen. On correlating the family history, the clinical features, oral findings and radiological investigations, the case was provisionally diagnosed as Marfan syndrome.

Images

Figure: 1

Figure: 2

Figure: 3
DISCUSSIONS

Marfan syndrome is one of the most common potentially lethal diseases inherited in Mendelian fashion. Characteristic features include progressive aortic dilatation associated with aortic valve incompetence, mitral valve prolapse and incompetence, lens dislocation and myopia, and a tall and thin body with long limbs, arachnodactyly, pectus deformities and sometimes scoliosis\(^7\). The diagnosis of a genetic disorder in a family and the possibility of testing for the disorder raise a number of issues. Involvement of genetics professionals (clinical geneticists and genetic counsellors) should be considered\(^6\). All family members potentially at risk should receive genetic counseling, life style modification advice and appropriate counseling should be given to the patient. Several diseases of the connective tissue disorder like Ehlers-Danlos syndrome, Shprintzen-Goldberg syndrome, Arterial tortuosity syndrome can be considered in the differential diagnosis of marfans syndrome. Some of the most complex diseases that show close resemblance to Marfans are MASS Syndrome, Loeys-Dietz syndrome.

Marfans is a rare hereditary connective tissue disorder which affects most parts of the body. Ghent nosology is used to establish the diagnosis in cases of marfans syndrome. Cardiovascular complications like aortic root dilatation and mitral valve prolapse are very common in marfans\(^9,10\). The mortality can be reduced by oral rehabilitation of the patient. Maintaining oral hygiene and palatal arch expansion can be carried out to improve the patients dental status of the individual. Regular cardiovascular, ocular and skeletal check up by means of echocardiography, MRI etc are highly recommended for the patients after the diagnosis of Marfans syndrome is established.

REFERENCES


