

Case Report

Marfan Syndrome- A case identified in dental clinicDr. Latika Bachani^{1*}, Dr. Ashok Lingappa², Dr. Shivaprasad S.³¹Post Graduate student, ²Prof and Head, ³Professor, Dept. of oral medicine and radiology, Bapuji dental college and hospital, Davangere-577004, Karnataka, India***Corresponding Author:**

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Abstract: Marfan syndrome is an autosomal dominant systemic disorder of the connective tissue. Children affected by this syndrome carry a mutation in one of the two copies of the gene encoding the protein fibrillin-1. It affects most organs and tissues, especially the skeleton, lungs, eyes, heart, and the large blood vessel that distributes blood from the heart to the rest of the body. A case report of Marfan syndrome has been reported which was diagnosed in the dental set-up.

Keywords: Marfan syndrome, Autosomal Dominant disorder, Oro-facial defects, Fibrillin-1

INTRODUCTION

Marfan Syndrome (MFS) is an autosomal disease of the connective tissue, which is characterized by skeletal anomalies and arachnodactyly. Many of its manifestations are misleading and are often interpreted as the common phenotypic traits of the general population, which sometimes makes it hard to diagnose in children. Some children have no signs or symptoms early in life but as they grow, they may develop the common Marfan traits. Herein we report a case of a 14-year-old female patient with unidentified Marfan syndrome.

CASE REPORT

A 16-year-old female patient reported to our department with a chief complaint of decay in her left lower back tooth since 1-2 years. History of food lodgment was present. No history of associated pain, sensitivity or discomfort was reported. Her past medical history revealed repeated episodes of hospitalization in her childhood due to weakness and recurrent fever. This was her first dental visit. Her mother revealed that her daughter had delay in the attainment of all the developmental milestones. She attained puberty at the age of 15 years and attained menopause after the first month. Her mother was of normal height and weight with unremarkable medical history. She had no siblings.

General examination revealed her to be tall (158 cms) (Fig.1), thin with elongated fingers and toes (arachnodactyly) (Fig.2). Her arm span length was 165 cms (Fig. 3). Characteristic Marfan bony changes were also present, in which wrist (Fig.4) and thumb signs (Fig.5) were noted, alongwith increased laxity of the

joints. She also suffered from blurred vision since birth. Blood pressure was recorded as 100/60 mmHg. On further evaluation, it was revealed that she has bilateral bluedot cataract. ECG was normal.

On extraoral examination (Fig.6), she was dolicocephalic with convex facial profile, wide-set eyes and incompetent lips. Intraoral examination revealed high arched palate (Fig.7), normal complement of permanent dentition and retained first quadrant deciduous second molar. 15 was palatally erupted. Class I caries was present in 16, 26, 36 and 46. She had End-on molar relation bilaterally with increased overjet.

On the basis of general physical examination and extraoral and intraoral findings, she was diagnosed with Marfan's syndrome and class I caries in 16, 26, 36 and 46.

A panoramic radiograph was taken which showed full complement of permanent dentition with presence of tooth buds of 18, 28, 38 and 48. Retained deciduous first quadrant second molar was seen. Coronal radiolucency involving enamel and dentin wrt 36 and 46 was present.

Cephalometric analysis verified maxillary prognathia (SNA = 88°), mandibular retrognathia (SNB = 78°), skeletal class II malocclusion (ANB = +10°), and a vertical growth pattern (PFH : AFH = 61.6%; Facial axis = 0°; Facial angle = 86°).

She was then referred for restorative treatment wrt class I caries followed by ophthalmic treatment.



Fig-1: Photograph depicting tall stature and elongated extremities



Fig-2: photograph depicting arachnodactyly



Fig-3: photograph depicting increased arm span length



Fig-4: Positive Wrist sign



Fig-5: Positive Thumb sign



Fig-6: Profile picture- Dolicocephalic



Fig-7: High arched palate

DISCUSSION

The National Marfan Foundation describes Marfan syndrome (MFS) as a heritable disorder of the connective tissue that can affect the heart, blood vessels, lungs, eyes, bones, and ligaments. Antoine Bernard Jean Marfan [1] first described MFS in 1896.

It is an autosomal dominant systemic disorder of connective tissues wherein the affected carries a mutation in one of the two copies of the gene that encodes the connective tissue protein fibrillin-1 (FBN 1), located at chromosome 15q-21.1 [2].

Recently, mutations in the transforming growth factor b-receptor 2 (TGFB2) genes on chromosome 3 and in the TGFBR1 gene on chromosome 9 were found in some families with apparent Marfan syndrome [3-5]. These 'Marfan syndrome type 2' [6] families seem less likely to have ectopia lentis.

The overall prevalence is 10.2 (95% CI, 9.8-10.7) per 100,000 individuals [6] and 26% of the cases have no family history [7].

Ocular, cardiovascular, and musculoskeletal abnormalities are considered the "classic triad" of MFS. It may involve other systems like the central nervous system, respiratory system, and skin. Patients with MFS typically show normal intelligence and cognitive development [9-12].

Affected individuals often are tall and slender, have arachnodactyly, scoliosis, and either a pectusexcavatum, pectuscarinatum, or ectopia lentis in eyes [13]. The incidence of mitral prolapse in such patients is essentially equal in children and adults of the same sex.

Common orofacial features include

dolichocephaly, enophthalmos, downward slanting palpebral fissures, malar hypoplasia, maxillary retrognathia (SNA < 80°), mandibular retrognathia (SNB < 78°), skeletal class II malocclusion, and hypermobility of the temporomandibular joint. Dentally, long narrow maloccluded teeth, large positive overjet, posterior crossbites, periodontal disease are also characteristic of MFS [9, 11, 14-21].

These patients many a times may face social stigmatization and discrimination that can reduce their quality of life and thereby lead to social withdrawal and psychiatric problems, especially during adolescence and young adulthood [22].

Diagnosis is mainly done using the Ghent criteria and a detailed clinical examination. The main criteria for diagnosis consist of clinical features that are typical of the syndrome and rarely occur in the general population. These include long limbs, scoliosis, pectuscarinatum, pectusexcavatum, ectopialentis, dilatation and/or dissection of ascending aorta, aortic regurgitation, and duralectasias. Minor criteria are present in individuals with the syndrome and often are seen in the general population. These include joint hypermobility, high palate, dolichocephaly, retrognathia, flat cornea, mitral valve prolapse, dilatation or dissection of the thoracic aorta, spontaneous pneumothorax, and recurrent hernias. In the presence of a non-significant family history, major criteria in at least two different organ systems and involvement of a third system are required. However, if evidence of a genetic mutation in the family is found, then one major criterion in an organ system and involvement of a second system are sufficient to establish a diagnosis of Marfan syndrome [11].

Differential diagnosis could include homocystinuria, familial aortic dissection, familial arachnodactyly, Ehlers-Danlos syndrome and multiple endocrine neoplasia IIB [23].

Although early diagnosis and refined medical and surgical management have increased median life expectancy from 40 to approximately 70 years, individuals with Marfan syndrome continue to suffer important morbidity [24]. The mean lifespan of untreated patients with MFS is approximately 32 years. Treated patients can expect to live beyond 70 years of age. Cardiovascular complications account for most premature mortality [25-27].

CONCLUSION

Marfan syndrome is the most common inherited connective tissue disorder with diverse clinical manifestations. This report underscores the importance of history taking and physical examination in a dental set-up that has helped in the diagnosis of unidentified Marfan syndrome case. Furthermore, the low life expectancy seen in the patients suffering from this

syndrome makes early diagnosis imperative to provide timely treatment and hence help in avoiding associated life threatening consequences. Therefore, a patient-centered team approach to address medical considerations and oral health is needed while acknowledging patient's psychosocial, behavioral, and intellectual abilities.

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