Case Report

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Marfan Syndrome: A Case Report and Review of Literature on Multipronged Approach

Dentistry Section

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ABSTRACT

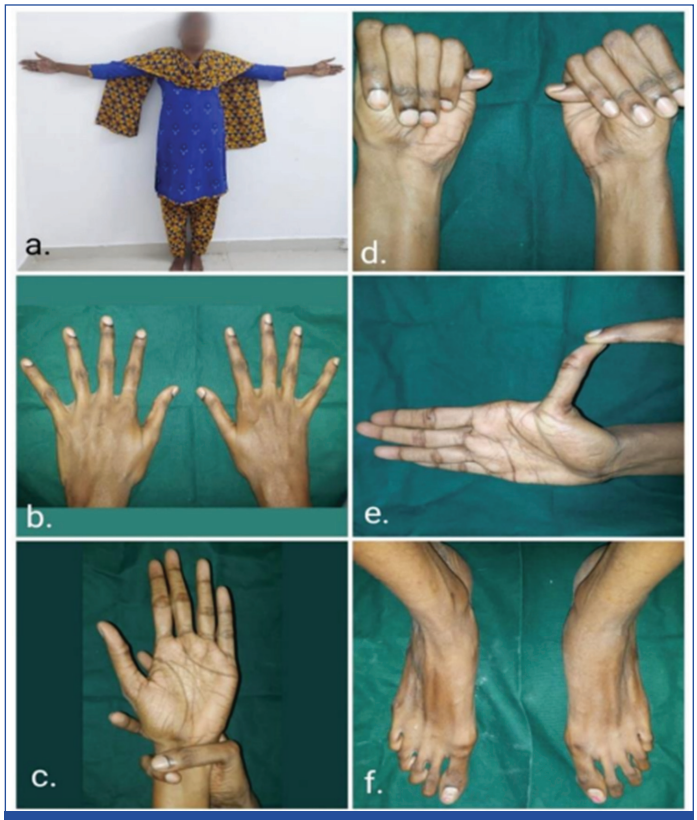
Marfan Syndrome (MFS) is a rare autosomal dominant inherited disorder of connective tissue due to the mutations in the Fibrillin-1(FBN1) gene located on chromosome 15q21.1. Alterations in this gene lead to widespread abnormalities affecting the ocular, cardiovascular, and skeletal systems, which are the characteristic features of the primary disease. Pulmonary, nervous system, cutaneous and oral cavity can also be affected in the secondary form of disease. Various oral manifestations of Marfan syndrome include bifid uvula, high arch palate, constricted arches, malocclusion and temporomandibular joint dysarthrosis. The most widely used diagnostic standard for Marfan syndrome is the revised Ghent nosology, which incorporates major and minor criteria developed through international expert consensus to ensure accurate identification of the disorder and to enhance patient management and outcomes. This paper discusses a 38-year-old female patient who reported with a complaint of pain in the right lower back tooth region, which had persisted for three weeks. Patient's history revealed that she had undergone mitral valve replacement surgery, sclerotomy for glaucoma and spinal fusion surgery for scoliosis. Management of the patient required a multidisciplinary approach integrating cardiology, ophthalmology, orthopaedics, and dental specialties to address this complex multisystem disorder. All dental surgical procedures requires antibiotic prophylaxis for the patients with Marfan syndrome who had undergone cardiac surgeries. Orthognathic surgery should be performed during early adolescent period for patients presenting with severe skeletal malocclusion.

Keywords: Fibrillin, Ghent nosology, Microfibril, Musculoskeletal

CASE REPORT

A 38-year-old female patient reported to Department of Oral Medicine and Radiology with a complaint of pain in the lower right back region of her teeth for the past three weeks. She was diagnosed with Marfan syndrome during her adolescent age and no other family members were affected by the disease. Genetic screening was not carried out and the diagnosis was based on the medical history and clinical findings. The patient had undergone mitral valve replacement surgery, spinal fusion surgery for scoliosis, sclerotomy for treatment of glaucoma and is under beta blockers and aspirin for the past 20 years. Patient also gives a dental history of restoration of 46 and extraction of 16 & 36 due to caries.

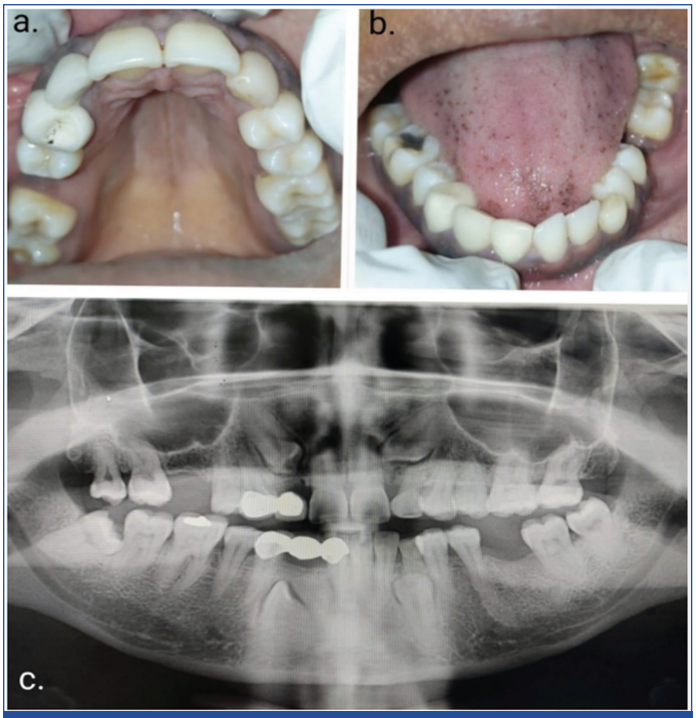
Physical examination revealed that the patient was tall and slender with an altered upper/lower segment ratio and increased arm height ratio (dolichostenomelia) [Table/Fig-1a], long, thin fingers (arachnodactyly) [Table/Fig-1b], and syndactyly. Wrist signs/ Walker Murdoch sign- overlapping of distal phalanges of thumb and fifth finger wrapped around the contralateral wrist [Table/ Fig-1c] and thumb sign/Steinberg sign- distal phalanx of thumb fully extends beyond ulnar border of hand and folded across the palm were positive [Table/Fig-1d]. The patient also had hyperextensibility of joints [Table/Fig-1e] and clawed toes [Table/ Fig-1f]. Her height was 178 cm which was greater than normal for Indian females. Orofacial features include dolichocephaly, malar hypoplasia, constricted maxillary and mandibular arches with a high-arched palate, and clinically missing 13,23,33,43,16,36 [Table/Fig-2a,b]. On radiographic examination, Orthopantomogram (OPG) revealed multiple impacted and retained deciduous teeth [Table/Fig-2c]. The treatment protocol which included removal of impacted teeth in relation to 48 and retained deciduous teeth in relation to 73, orthodontic traction of impacted teeth along with replacement of missing teeth with implants under antibiotic coverage was advised. The patient did not report for treatment or any follow-up visits.



[Table/Fig-1]: a) Dolichostenomelia; b) Arachnodactyly; c) Thumb sign/Steinberg sign; d) Positive Wrist signs/Walker Murdoch sign; e) Hyperextensibility of joints; f) Clawed toes.

DISCUSSION

Marfan syndrome is an autosomal dominant disorder of connective tissue primarily characterised with anomalies affecting the musculoskeletal system, the cardiovascular system and the eyes. The primary gene defect in MFS lies on chromosome 15q21.1, within the coding region of the FBN1 gene. FBN1 is a large 350



[Table/Fig-2]: a,b) Constricted maxillary and mandibular arch with high arched palate; c) OPG reveals multiple impacted teeth and retained deciduous.

with dislocation in the superotemporal quadrant in both eyes [4]. Folkestad L et al., did a nationwide register-based cohort study and concluded that the patients with MFS have altered bone geometry and bone microstructure with lower bone mass, leading to age related bone loss, and an increased risk of fractures. In the present case reported, the patient presented with cardiovascular, ocular, musculoskeletal and dental abnormalities [5].

The diagnosis of Marfan syndrome is usually challenging, Revised Ghent nosology scoring [6] helps to arrive at the diagnosis, especially in cases where there is no family history or genetic screening. In the present case, score was 7 with positive features of wrist and thumb sign, reduced upper segment/lower segment ratio and increased arm/height. Scoliosis or thoracolumbar kyphosis, facial features (dolichocephaly, malar hypoplasia, retrognathia), and mitral valve prolapse. It is essential to rule out other syndromes with overlapping features which is listed in [Table/Fig-3] [6].

Management of patients with MFS having severe cardiovascular system, ocular disorders and musculoskeletal system usually require immediate surgical intervention to prevent the severity and disease progression [Table/Fig-4] [7-13]. Surgical repair of mitral valve prolapse, spinal surgery for scoliosis and sclerotomy for treatment of glaucoma was done in this case. Orofacial manifestations like dolichocephaly, malar hypoplasia, Gothic arch-like palate and malocclusion may require orthognathic surgeries or orthodontic treatment with palatal expanders. Early intervention

Differential diagnosis	Gene	Discriminating features
Loeys-Dietz Syndrome (LDS)	TGFBR1/2	Bifid uvula/cleft palate, arterial tortuosity, hypertelorism, diffuse aortic and arterial aneurysms, craniosynostosis, clubfoot, cervical spine instability, thin and velvety skin, easy bruising
Shprintzen-Goldberg syndrome (SGS)	FBN1 and other	Craniosynostosis, mental retardation
Congenital contractural arachnodactyly (CCA)	FBN2	Crumpled ears, contractures
Weill-Marchesani syndrome (WMS)	FBN1 and ADAMTS10	Microspherophakia, brachydactyly, joint stiffness
Ectopialentis Syndrome (ELS)	FBN1, LTBP2, ADAMTSL4	Lack of aortic root dilatation
Homocystinuria	CBS	Thrombosis, mental retardation
Familial Thoracic Aortic Aneurysm syndrome (FTAA)	TGFBR1/2, ACTA2	Lack of Marfanoid skeletal features, levido reticularis, iris flocculi
FTAA with Bicuspid Aortic valve (BAV)	-	-
FTAA with Patent Ductus Arteriosus (PDA)	MYH11	-
Arterial Tortuosity Syndrome (ATS)	SLC2A10	Generalised arterial tortuosity, arterial stenosis, facial dysmorphism
Ehlers-Danlos syndrome (vascular, valvular, kyphoscoliotic type)	COL3A1, COL1A2, PLOD1	Middle sized artery aneurysm, severe valvular insufficiency, translucent skin, dystrophic scars, facial characteristics

[Table/Fig-3]: Features of differential diagnosis [6].

kDa glycoprotein, produced and secreted by fibroblasts, and incorporated into the Extracellular Matrix (ECM) as insoluble microfibrils. These microfibrils serve as a scaffold for the deposition of elastin that are necessary to build proper elastic architecture to provide elasticity to dynamic connective tissues. Structurally, each FBN1 monomer contains multiple Epidermal Growth Factor (EGF). Mutations in FBN1 resulted in weak and disordered elastic fiber formation, causing a disruption of the microfibril network connections to the adjacent interstitial cells [1].

Majority of the patients affected by Marfan syndrome present with serious cardiovascular system abnormalities and ocular disorder which may require surgical intervention, but it may affect other systems also. In a case reported by Yanamadala PK et al., patient initially presented with complaints of chest pain, shortness of breath, and joint laxity. Echocardiography showed severe aortic root dilation and mitral valve prolapse, requiring immediate medical attention [2]. Encarnación-Santos DA et al., reported a case with a sudden onset of severe headache followed by a gradual loss of consciousness. His medical history was significant for Marfan syndrome. They found a spontaneous subdural hematoma in the left frontoparietotemporal area [3]. Upasani DS et al., reported a case with complaints of diminished visual acuity in both eyes since birth. The patient exhibited cataractous changes in the lens

of skeletal deformities may prevent the well-formed malocclusion, but the patient did not undergo any such treatment. Patient was advised extraction of 48 under antibiotic cover and orthodontic

System	Manifestations	Treatment
Musculoskeletal [7]	➢ Thoracolumbar scoliosis/Kyphosis ➢ Pectus excavatum ➢ Pectus craniatum ➢ Hindfoot Valgus ➢ Arachnodactyly ➢ Dolichostenomelia ➢ Hyperextensibility of joints ➢ Dolichocephaly ➢ Enophthalmos ➢ Down slanting palpebral fissures ➢ Malar hypoplasia ➢ Mandibular retrognathia	➢ Bracing, spinal fusion surgery ➢ Vaccum bell, Open Ravitch/ Nuss Procedure ➢ Chestbracing, Ravitch ➢ Chestbracing, Ravitch Procedure
Cardiac [8]	➢ Mitral valve prolapse ➢ Aortic dilatation ➢ Aortic regurgitation ➢ Aneurysms	➢ Surgical repair of mitral valve ➢ Surgery(replacement of aortic root with a composite graft) ➢ Beta blockers/Angiotensin receptor blockers/Aortic valve repair surgery ➢ Open surgical repair/ Endovascular repair

Ocular [9]	➢ Myopia ➢ Cataracts ➢ Retinal detachment ➢ Ectopia lentis ➢ Glaucoma	➢ Glasses/contact lenses ➢ Laser assisted cataract surgery ➢ Scleral buckling and vitreous Surgery ➢ Eye glasses/Lens extraction and Artificial intraocular lens replacement ➢ Antiglaucoma medications ➢ Minimally invasive glaucoma surgeries (or) Non-penetration deep sclerotomy
Pulmonary [10]	➢ Pneumothorax	➢ Conservative treatment (oxygen by facemask and rest) ➢ Surgery {Video-Assisted Thoracoscopic Surgery (VATS) for bullectomy and pleurdesis without perioperative complications}
Dental manifestations [11]	➢ High arched palate, ➢ Crowding, and ➢ Posterior openbite.	➢ Orthodontic treatment
Nervous [12]	➢ Dural ectasia	➢ Sufficient hydration and bed rest/epidural blood patch, continuous epidural saline infusion, or an epidural injection of fibrin glue ➢ Surgical repair
Cutaneous [13]	➢ Striae atrophicae	➢ Tretinoin cream ➢ Light and laser therapies ➢ Radiofrequency energy devices

[Table/Fig-4]: Clinical manifestations and management of Marfan syndrome [7-13].

traction of impacted teeth in relation to 13, 23, 33, and 43 for which the patient did not report back. Generally, antibiotic prophylaxis is recommended for dental procedure which involves the gingival tissue, the periapical tooth area, or biopsy/perforation of the oral mucosal tissue [14].

There are significant advancements in MFS treatment over the past few decades, substantial work remains to be done. Current drug treatments for MFS do not provide a cure; instead, they primarily aim to slow the progression of disease. As a result, creating more effective therapeutic approaches continues to be an important focus of ongoing research which includes genome editing technology, Adeno-associated Virus-mediated gene therapy, and induced Pluripotent Stem Cells (iPSCs) [15].

CONCLUSION(S)

The early diagnosis and medical management of Marfan syndrome is very important, as they increase patient's quality of life. Dental treatments can be carried out safely by focusing on precautions that could avoid cardiovascular complications. Some individuals with this syndrome may not need treatment, but all patients are advised to have regular dental visits.

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