Marfan’s Syndrome: An Orthodontic Perspective

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ABSTRACT

Marfan syndrome is a heritable disorder of connective tissue that can affect the heart, blood vessels, lungs, eyes, bones and ligaments. It is characterized by tall stature, elongated extremities, scoliosis and a protruded or caved-in breastbone. Patients typically have a long, narrow face. A high-arched palate produced by a narrow maxilla and skeletal Class II malocclusion due to mandibular retrognathia are other common features. For a patient with no family history of this disorder, at least three body systems must be affected before a diagnosis can be made—skeletal, cardiovascular and ocular. Individuals affected by the syndrome routinely demand orthodontic treatment to correct the orofacial manifestations. This case report presents two patients who reported to the department of orthodontics with a chief complaint of protruding upper anteriors, but on thorough clinical examination and investigation were diagnosed as Marfan syndrome. A brief overview of the challenges involved in treating such patients is discussed herewith.

Keywords: Marfan’s, Syndrome, Malocclusion.

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INTRODUCTION

The National Marfan Foundation (NMF) describes Marfan’s syndrome as a heritable disorder of connective tissue that can affect the heart, blood vessels, lungs, eyes, bones and ligaments. The condition was named after a French pediatrician, Antoine Bernard-Jean Marfan, who first described its occurrence (1896) in a 5-year-old girl named Gabrielle with ‘spider’s legs’ or dolicostenomely (from the Greek word stenos—narrow, slender; melos—limb); the patient was noted to have disproportionately long and thin arms, legs, fingers and toes. Marfan’s syndrome is an autosomal dominant genetic disorder of the connective tissue. It is classified as type I or type II.

Marfan Type I or Classic Marfan’s Syndrome

Males and females are equally affected. Mutations in the FBN1 gene located on chromosome 15 causes type I Marfan’s syndrome. The fibrillin-1 glycoprotein encoded by the FBN1 gene is required for the formation of elastic fibers in connective tissue. A mutation in the gene can decrease the quantity and quality of fibrillin-1. This in turn can lead to weakened structural support, especially in areas where elastic fibers are found in abundance. Consequently, the aorta, ligaments and ocular muscles are among the most frequently affected parts of the body.

Type II Marfan’s Syndrome

It is less common and is due to a mutation in the gene that encodes transforming growth factor beta receptor 2 (TGFBR2). The protein synthesized by this gene transmits signals from the cell surface to the nucleus, thereby affecting cell division and growth. The clinical presentation of type II Marfan’s syndrome resembles that of classic Marfan’s syndrome, with the exception that the ocular system usually is not involved.

Symptoms

Marfan’s syndrome affects different people in different ways. Symptoms range from mild-to-severe and they progress with age in most cases. Skeletal abnormalities are the most readily visible signs. Affected individuals are markedly taller than their age-matched unaffected counterparts. Long slender limbs, fingers and toes are characteristic. Arms are exceptionally long and their expanse when outstretched is often greater than the height of the individual. Other typical features include abnormal curvature of the spine (scoliosis), caved-in (pectus excavatum) or protruding sternum (pectus carinatum) and abnormal joint flexibility.

Diagnosis

Marfan’s syndrome is diagnosed primarily on clinical grounds. Imaging studies, such as radiography, echocardiography and
magnetic resonance imaging (MRI) facilitate detection and monitoring of cardiovascular diseases.³,⁸

CASE REPORTS

Case 1

A 17-year-old boy presented with a chief complaint of irregular upper anterior teeth. Medical history was not significant. Complained of fatigue and palpitations on daily working.

General Examination

Patient had a long thin body built with a height of 6 feet 3 inches and arm span of 6 feet 7 inches (Fig. 1), spidery fingers (Fig. 2), wrist sign positive and thumb test positive.

On extraoral examination: Patient had a dolichocephalic skull form, leptoprosoptic face, a convex facial profile (Figs 3A to D) and malar hypoplasia but a normal lower facial height. His lips were incompetent, protrusive and upper lip short but the nasolabial angle was normal.

Intraoral examination: It revealed a permanent dentition with a poor oral hygiene. Malocclusion was Angle class II molar and canine relationship, with a high-arched palate and crowding (Figs 4A to E) in upper anteriors.⁹

Functional examination: It indicated normal temporo-mandibular joint (TMJ) and mandibular movements.

Suspecting Marfan’s syndrome the patient was referred to a physician for systemic examination.

Systemic Examination

On skeletal examination, patient had a scoliosis (Fig. 5), dolichostenomalia, pectus carinatum (Fig. 6), archnodactyly (Fig. 7) and retrognathia mandible and was conformed on PA chest radiograph, lateral chest radiograph, radiograph of femur, hand-wrist radiograph and lateral skull radiograph.

CVS examination: It revealed tachycardia and mitral valve prolapse on ECG and 2D echocardiogram.
Ophthalmologic examination: It reported myopia, ectopic lentis, upward gazing of eye ball.

With the above all findings, the diagnosis was type 1 Marfan’s syndrome.

Case 2

A 17-year-old boy presented with a chief complaint of proclined upper anterior teeth. Complained of knee joint pain. Familial history of tallness reported.

General Examination

Patient had a long thin body built with a height of 6 feet 2 inches and arm span of 6 feet 4 inches (Fig. 8), spidery finger, thumb test positive and wrist sign positive.

On extraoral examination: Patient had a dolichocephalic skull form, leptoprosopic face, a convex facial profile (Figs 9A to D). His lips were incompetent, protrusive and upper lip short. The mentolabial fold was abnormally accentuated and malar hypoplasia. Excessive gingival display on smiling and increased lower face height. There was a mild facial asymmetry deviated toward right. His nose and alar bones deviated to the right with prominent nasal hump.

Intraoral examination: Poor oral hygiene with generalized gingival inflammation, root stumps of 36 and carious 31 (Figs 10A to E). Malocclusion was Angle Class II division 1, with a high-arched palate, proclined upper anteriors but a prominent finding in this patient was the presence of parastyle on 16,13.

Functional examination: TMJ movement was normal. On opening and closing movements, mandible deviated toward left.
Systemic examination was carried out by physician and was referred to a cardiologist, ophthalmologist and radiologist.

**Systemic Examination**

On skeletal examination, patient had scoliosis (Fig. 11), dolichostenomelia, pectus carinatum (Fig. 12), archnodactyly (Fig. 13) and retrognathic mandible (Fig. 14) and was confirmed on the PA chest radiograph, lateral chest radiograph, radiograph of femur, hand-wrist radiograph and lateral skull radiograph in Government Medical College and Hospital, Nagpur.

*CVS examination:* It revealed Austin flint murmur and aortic dilation.

Ophthalmologic examination reported no obvious finding.

With the above all findings, the diagnosis was type 2 Marfan’s syndrome.

Since, both the patients had cardiac system involvement and as per the American Heart Association they fall in moderate risk group they are under the care of cardiologist.

**Orthodontic Consideration in Marfan’s Syndrome**

- Ascertain cardiac condition—establishment of the risk of endocarditis and consultation with cardiologist.
Degree of oral hygiene must be established. No orthodontic treatment to be started unless there is excellent oral health.
When patient is at risk, prior to any orthodontic treatment procedure a 0.2% chlorohexidine mouthwash may be used.
Antibiotic prophylaxis should be used where required or for procedures that may cause gingival bleeding.
Patient should give a clear commitment to maintain a very high standard of oral cleanliness.
Informed consent—the patient and family to be fully involved in the consent process.
Periodic periodontal consultation.
Efforts should be made to avoid any form of gingival or mucosal irritation.
Bondable brackets and tubes should be used rather than band material.
Orthodontist should be particularly vigilant for any deterioration in gingival health.
Special attention should be given about positioning of tubes and hooks.
Avoid fixed acrylic appliances like rapid maxillary expansion and nance button.
Apply light force for retraction.
Duration of treatment should be minimal.
When possible nonextraction approach should be followed.
Superalastic wires should be used as it require less frequent wire adjustment.
Elastic ligatures should be used instead of stainless steel wire.
Proper measures should be taken to control infection.
Duration of retention must be longer because longer duration required for collagen fiber organization.

HYPOTHESIS AND FACT

Then the question is to treat or not to treat these patient?
First hypothesis: Connective tissue disorders often are associated with extensive periodontal tissue breakdown.

Second hypothesis: Marfan’s syndrome is associated with severe periodontitis.

With the development of molecular biology, the fact about Marfan’s syndrome is very clear, FBN1 gene has least effect on the periodontal ligament because it has least effect on type 1 collagen which is the main constituent of periodontal ligament.

In type 2, transforming growth factor beta receptor 2 (TGFBR2) plays an important role in Marfan’s syndrome. TGFBR2 is least associated with periodontium.

CONCLUSION

Orthodontic treatment is feasible in Marfan’s syndrome but special precautions need to be taken. These include medical consultation, informed consent and oral hygiene maintenance. The orthodontist should aim for an esthetically acceptable and functional result but not necessarily for orthodontic perfection.

REFERENCES