Marfan Syndrome: A Report of Two Cases and Review

Vinay Kumar Reddy K, Venkata Satya Ramesh T, Koteeswaran D, Jayakiran M, Sivaranjani Y

1Professor and Head, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
2Senior Lecturer, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
3Professor, Department of Oral Medicine and Radiology, Meenakshi Ammal Dental College and Hospital, Chennai, Tamil Nadu, India
4Senior Lecturer, Department of Oral and Maxillofacial Pathology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India
5Reader, Department of Oral and Maxillofacial Pathology, Mamata Dental College and Hospital, Khammam, Andhra Pradesh, India

Correspondence: Vinay Kumar Reddy K, Professor and Head, Department of Oral Medicine and Radiology, Mamata Dental College and Hospital, Staff Quarters, Penna G-4 Mamata Hospital Campus, Giriprasad Nagar, Khammam, Andhra Pradesh, India, Phone: 9849052434, e-mail: kundoorvinayreddy@yahoo.in

ABSTRACT

Marfan syndrome is an autosomal dominant disorder of the connective tissues, resulting in abnormalities of the musculoskeletal system, cardiovascular system and eyes. It has a prevalence of 1 in 100,000 population and occurs in all ethnic groups. It may be familial or due to new mutation (30%), in the fibrillin gene on arm of chromosome 15. It is estimated that one person in every 3000 to 5000 has Marfan’s syndrome may have cardiovascular abnormalities and may be complicated by infective endocarditis. About 90% of Marfan patients will develop cardiac complications.

Keywords: Marfan syndrome, Autosomal dominant, Arachnodactyly, Pectus excavatum, Mitral valve prolapsed, Myopia, Infective endocarditis.

INTRODUCTION

Marfan syndrome is an inherited autosomal dominant disorder of the connective tissue that causes abnormalities of the patient’s musculoskeletal system, cardiovascular system and eye. It is associated with the mutation in the fibrillin-1 gene located on chromosome 15 (locus 15q, 21,1). It is named for the French pediatrician, Antoine Marfan (1858-1942), who first described it in 1896. Marfan syndrome is sometimes called arachnodactyly, which means ‘spider-like fingers’ in Greek, since one of the characteristic signs of the disease is disproportionately long fingers and toe. It is estimated that one person in every 3000 to 5000 has Marfan syndrome, which can be complicated by cardiovascular abnormalities and further can be complicated by infective endocarditis.

CASE REPORT

Case 1

A 24-year-old male patient, Mr Kumar, came to our Department of Oral Medicine and Radiology, Meenakshi Ammal Dental College, Chennai with a chief complaint of decayed tooth with pain in the lower left back tooth region.

Medical history revealed that the patient was having myopia in both the eyes and mitral valve prolapsed. Family history revealed that both his mother and brother were diagnosed with Marfan syndrome.

General examination of the patient revealed long and narrow skull, tall stature (175 cm), arm span > total height (190 > 175 cm), pectus excavatum, reduced upper and lower segment ratio (Figs 1 and 2). Arms, legs, fingers and toes are disproportionately long in relation to the rest of the body.

Fig. 1: Long and narrow skull
Marfan Syndrome: A Report of Two Cases and Review

JIAOMR

composed of 65 exons on chromosome 15q15-q21. Generally, it is inherited as an autosomal dominant trait, which implies that only one parent need have the errant gene and that each child with one affected parent has a 50-50 chance of inheriting the disorder. In 25 to 33% of the cases, however, neither parent has the disorder, but instead the syndrome develops because of a spontaneous mutation in either the egg or the sperm.6

MFS is an inherited, degenerative connective tissue disorder with skeletal, ocular, ligamentous, cutaneous, pulmonary, neurologic, abdominal and cardiovascular manifestations. 7,8 Skeletal manifestations include tall stature, thin body build, long arms and legs (dolichostenomelia), long fingers and toes (arachnodactyly), hyperextensibility, pectus deformity, scoliosis, joint contractures and narrow, high arched palate. Both of our cases showed arachnodactyly and dolichostenomelia. The sternum may either protrude, giving a pigeon-breast appearance (i.e. pectus carinatum) or indent, giving a funnel chest appearance (i.e. pectus excavatum). Pectus excavatum was seen in both of our cases. Abnormal spinal curvatures are also common: Kyphosis (30 to 35% of patients), scoliosis, thoracic lordosis, a straight thoracic spine and combined scoliosis and kyphosis (i.e. kyphoscoliosis).

Case 2

A 52-year-old male came to the department of oral medicine and radiology with a chief complaint of pain in the lower left back tooth region, for past 20 days. Medical history revealed that the patient has underwent surgical treatment for mitral valve prolapse one year ago and also revealed that the patient was having myopia in both his eyes.

General examination of the patient revealed long and narrow skull, tall stature (182 cm), arm span > total height (194 > 182 cm), pectus excavatum, and a surgical scar was present in the chest region (Fig. 5). Arms, legs, fingers, and toes are disproportionately long in relation to the rest of the body (Fig. 6). His vital signs were within the normal limits. Intraoral examination revealed high arched palate.

Considering the medical and surgical history, clinical and laboratory findings, diagnosis of Marfan syndrome was made.

DISCUSSION

Marfan syndrome (MFS), first described in a 5-year-old child by the French pediatrician Antoine Marfan in 1896, is a dominantly inherited connective tissue disease with a wide range of phenotype severity in the skeletal, ocular and cardiovascular systems.3 Marfan syndrome is a spectrum of disorders caused by a heritable genetic defect of connective tissue that has an autosomal dominant mode of transmission.4

MFS is a heritable disorder where abnormalities of fibrillin protein are encoded by the fibrillin-1 gene (FBN1), a large gene...

Fig. 2: Tall stature and pectus excavatum

Fig. 3: Arachnodactyly of fingers

Fig. 4: Arachnodactyly of toes

(Figs 3 and 4). His vital signs were within the normal limits. Intraoral examination revealed high arched palate, generalized enamel hypoplasia.

Investigations were done which showed normal complete hemogram, and echocardiogram was done which showed mitral valve prolapse with trivial mitral regurgitation, mild aortic regurgitation and tricuspid valve prolapse.

Considering all the clinical and laboratory findings, diagnosis of Marfan syndrome with mitral valve prolapse was made.
Cardiovascular manifestations include mitral valve prolapse, mitral valve regurgitation, aortic dilatation, aortic regurgitation, acute aortic aneurysm with or without regurgitation, tricuspid valve and pulmonary valve regurgitation. Between 75 and 85% of Marfan patients have loose or ‘floppy’ mitral valves, which are the valves that separate the chambers of the heart. Mitral disorders tend to present early in life, whereas aortic involvement progresses with age and generally presents in adolescence and adulthood. Interestingly, mitral disorders are seen more frequently among women, while aortic dilatation and aortic regurgitation are common among men. Both of our cases showed mitral valve prolapse and echocardiogram of case one showed mild aortic regurgitation and tricuspid valve prolapse. Case one was referred to cardiologist for the management of mitral valve prolapse.

Ocular manifestations include myopia, tremor of the iris with eye movement (i.e. iridodonesis), glaucoma, cataracts and spontaneous retinal detachment. Myopia was present in both case one and two. The ligaments and tendons are often weak and lax, with hyperextensibility of the small joints. For example, a patient with MFS can usually pull the thumb back, so that it touches the radius and flex the thumb across the palm so that it extends beyond the ulnar side of the hand when the fingers are flexed in a fist. Ligament and tendon laxity may produce genu recurvatum, along with pes planus or pronation of the feet or both. Those with MFS often develop stretch marks (striae atrophicae) without significant weight changes. The marks often appear in sites subject to stress, including the shoulders, hips and lower back. Also, subcutaneous fat is sparse.

Those with MFS often show a stretching of the dura mater, particularly in the lower portion of the spine. This enlargement (dural ectasia) gradually pushes on the vertebrae and wears them down, enlarging the canal and thinning the vertebrae.

Fig. 5: Tall stature and long narrow skull of the patient

A predisposition to hernias can occur in childhood or later in life.

According to Baden and Spirgi, who have reviewed the oral manifestations of this disease, a high arched palatal vault is very prevalent and may be a constant finding. Bifid uvula and malocclusion are also reported. In addition, multiple odontogenic keratoceysts of the maxilla and mandible have occasionally reported by Oatis et al. One additional finding that sometime present is temporomandibular dysarthrosis. Both of our cases showed high arched palate and malocclusion. Skull radiographs (anterioposterior views and lateral views) may demonstrate a high arched palate, increased skull height and enlarged frontal sinus.

The diagnosis of MFS with classical phenotype remains largely clinical. The diagnosis is made by taking a family history and thorough examination of the patient’s eyes, heart and bone structure. The examination should include an echocardiogram taken by cardiologist, a slit-lamp eye examination by an ophthalmologist, and a work-up of the patient’s spinal column by orthopedic specialist. New investigation is mutation analysis (study of FBN1 gene).

The major goal in managing a patient with MFS is to prevent life-threatening complications through early diagnosis and treatment. While there is no cure for the disorder, careful management can improve the prognosis and lengthen the lifespan. Ideally, those with MFS should be treated by a physician familiar with the condition and its effects on all body systems. This physician can coordinate referrals to specialists and counsel on prognosis and treatment options.

The prognosis for patients with Marfan has improved markedly in recent years. As of 1995, the life expectancy of people with the syndrome has increased to 72 years, up from 48 years in 1972. This dramatic improvement is attributed to new surgical techniques, improved diagnosis and new techniques of medical treatment. The most important single factor in improving the patient’s prognosis is early diagnosis. The earlier that a patient can benefit from the new techniques
and lifestyle modifications, the more likely he or she is to have a longer life expectancy.

**CONCLUSION**

MFS can present with varying degrees of expression and penetrance; consequently, not all those with MFS present with the same phenotype. So, a thorough clinical history of the patient and laboratory evaluation should be done for early diagnosis of the condition. The early diagnosis will increase the life expectancy of the patient with this syndrome.

**REFERENCES**


