Journal of Cardiovascular Disease Research

ISSN: 0975-3583, 0976-2833 VOL12, ISSUE02, 2021

Saga of multiple teeth – Case report of Marfan Syndrome

Dr. Bhavika Vhatkar
Associate Professor, Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email-id – drpolvhatkar@gmail.com
Dr. Naveenkumar Shetty
Professor, Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email-id -nksrad@rediffmail.com
Dr. Hemant Bhutani
Lecturer, Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email-id-drhemantbhutani@gmail.com
Dr. Vagisha Choudhary
Post Graduate Student, Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email id – vagishac14@gmail.com
Dr. Janhavi Chandure
Post Graduate Student
Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email id- janvichandure96@gmail.com
Dr. Sagar kumar
Post Graduate Student, Department of Oral Medicine and Radiology
D Y Patil University, School of Dentistry, Navi Mumbai
Email Id – sagar5115@gmail.com

ABSTRACT :
Marfan syndrome is a rare autosomal dominant disorder of the connective tissue, which shows skeletal, ligamentous, oro-oculofacial, pulmonary, neurological manifestations. The incidence of the syndrome is approximately 1:5000. There is no reported difference based on the gender, ethnicity and geographic groups. Marfan Syndrome is caused by pathogenic variants of *FBN1gene*, which encodes a large protein fibrillin-1, which is a major structural component of the extracellular matrix . The diagnosis of the syndrome becomes difficult as it is essentially a clinical one, relying mainly on family history, physical examination and investigation of involved organ systems. Marfan syndrome is mostly accounted by the dentists at an early age. The present case illustrates the typical features of Marfan syndrome and its treatment carried out .
Key Words: Marfan syndrome, FBN1 gene, arachnodactyly

INTRODUCTION :

Marfan syndrome is a rare autosomal dominant genetic disorder caused by a mutation on the gene encoding fibrillin-1.1 The gene is located on chromosome 15 (15q21) (6, 7).1 The prevalence of this is 1 in 5–10,000 individuals.2 Almost 92% of patients with Marfan's

ISSN: 0975-3583, 0976-2833 VOL12, ISSUE02, 2021

syndrome show mutation on FBN1 gene.1 The diagnosis of Marfan syndrome relies on a clinical diagnostic criteria known as revised Ghent score. The individuals with Marfan syndrome appear tall, thin, with chest and ribs protruding outwards that is known as pectus carinatum, and have long, thin extremities and unusually long, spidery digits, and scoliosis.1 Due to lack of specific laboratory tools, the diagnosis depends upon the Ghent criteria, major and minor diagnostic features of the syndrome. Dental professionals need to be well-versed in both the orofacial and physical characteristics of Marfan syndrome in order to make a preliminary diagnosis of their patients and promptly send them to experts for further therapy.3

CASE REPORT :

A 17 year old male patient visited the department of Oral Medicine and Radiology with a chief complaint of missing upper front tooth since childhood.

Patient gave no relevant medical history. The family history revealed of parents having a consanguineous marriage. The mother also revealed that his elder sister had similar features to him The patient's social and intellectual development was normal.The patient appeared too thin and slender with his body weight being less than average for his age and sex.

On physical examination his showed elongated fingers with thickening at the phalange joints (figure 1). He had disproportionately long arms and legs when compared to the trunk. His arm span was about 3.5inches more when compared to his height (figure 2). The patient had positive Walker Murdoch wrist sign which means on wrapping the distal phalanx and little finger around the opposite wrist there is overlapping of complete distal phalanx and little finger (figure 3A) and a positive Steinberg thumb sign, which means on folding the thumb into fist the distal phalanx extends beyond the ulnar border (figure 3B) indicating extreme ligament laxity. An examination of the feet revealed flat feet with elongated toes.

FIGURE 1 :showing arachnodactyly which means elongated fingers

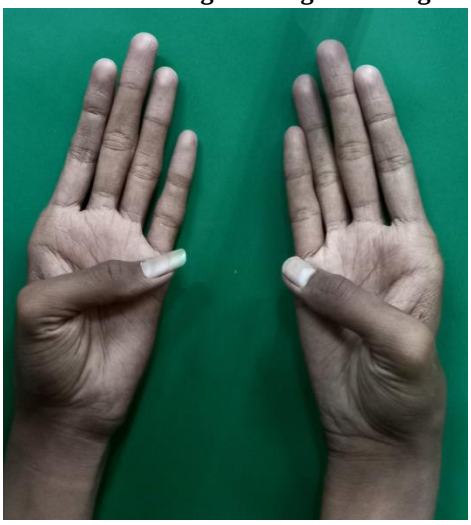

FIGURE 2 : showing increased arm pan compared to height

FIGURE 3A : showing positive

FIGURE 3B : showing positive Walker Murdoch wrist

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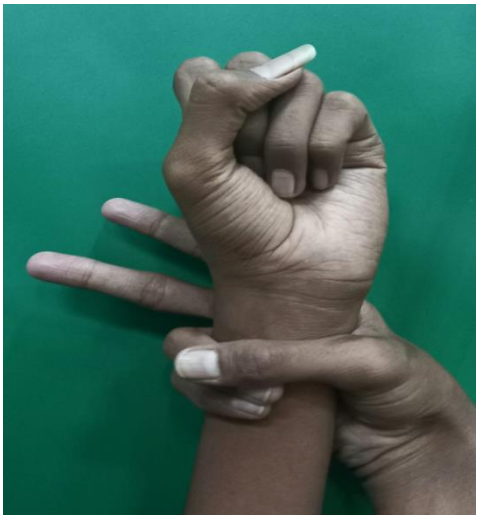
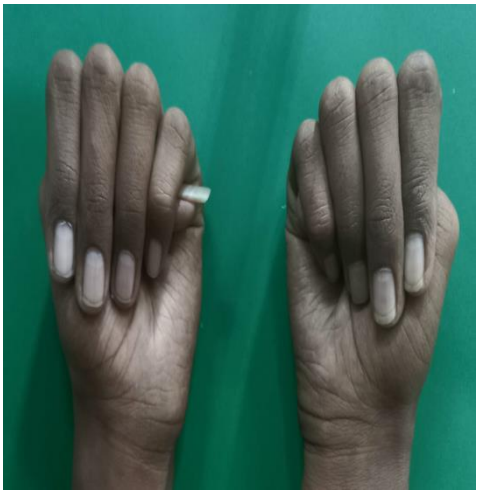




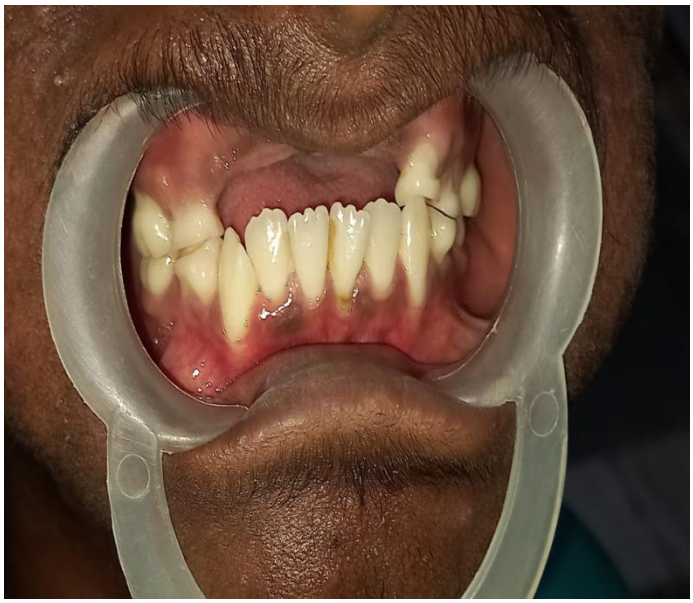
sign **Steinberg thumb sign**

Extraoral examination revealed narrow cranium with dolicocephaly and leptoprosopic facial feature. He also showed evident malar hypoplasia and downwards slanting palpebral fissures. The patient also showed 2 discrete swellings on the right side of the neck and multiple sebaceous swelling on the face.

The intraoral findings included high arch palate, multiple missing permanent teeth.

The teeth present clinically were 54, 55, 63, 64, 65, 74, 75, 84, 85, 31, 32, 33, 41, 42, 43. (figure 4). He also had cervical abrasion and gingival recession in relation to 54, 55, 64, 65.The boy had poor oral hygiene.

FIGURE 4 : showing intraoral image of missing tooth and retained deciduous teeth



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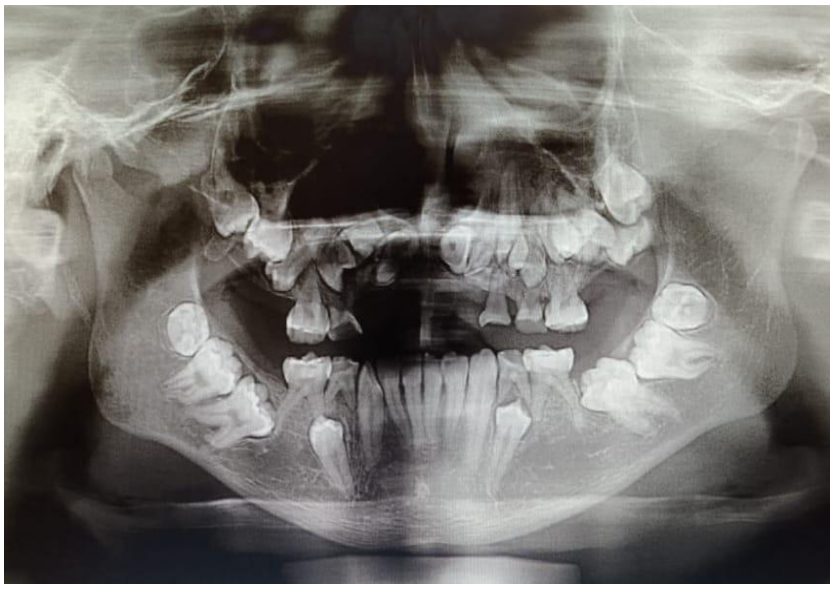
INVESTIGATIONS :

The patient was advised OPG , blood test , serum calcium and serum alkaline phosphatase level. Though the blood test and serum calcium and serum alkaline phosphatase level was normal. The orthopantomogram revealed multiple impacted permanent teeth which included 11, 12, 13, 14, 15, 16, 17, 21, 22, 23, 24, 25, 26, 27, 28, 34, 36, 37, 38, 44, 46, 47, 48 and erupted 54, 55, 63, 64, 65, 74, 75, 84, 85, 31, 32, 33, 41, 42, 43 (figure 5).

FIGURE 5: showing OPG with multiple impacted teeth



DIFFERENTIAL DIAGNOSIS :

A differential diagnosis of Gardener's syndrome was made. Other differential diagnosis including Noonan syndrome and homocystinuria was made as homocystinuria has similar features as pectus carinatum being common.[3]

TREATMENT :

Oral prophylaxis was performed for the patient. A multidisciplinary treatment was decided for the patient consulting the paediatric dentist, orthodontist and a prosthodontist. It was decided to perform tooth supported full mouth rehabilitation for the patient, thus not hampering the impacted tooth and the alveolar bone width.

DISCUSSION :

Marfan syndrome was first described by a pediatrician Antoine BernardJean Marfan in 1896.[4] Marfan syndrome is a hereditary disorder affecting multiple organs and systems. It occurs due the mutation of fibrillin protein on gene FBN1 .

Even among members of the same family, the clinical presentation and severity of the illness might differ. Patients' life opportunities are limited due to physical impairment and cosmetic difficulties, which causes frustration and low self-esteem.[4] The majority of Marfan syndrome

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sufferers do not have all of the defining characteristics and/or problems of the condition. Sometimes, parents may not notice a child's ailment because it is asymptomatic.[1]

Over the years, a diagnosis has frequently depended on the treating physician's ability to identify the evidence of numerous clinical symptoms dispersed over multiple organ systems. Even though the majority of patients do exhibit these common clinical symptoms, the genetic defect's varied penetrance causes inconsistent onset timings, which can often make a diagnosis challenging. The syndrome primarily involves the malformations of skeletal, cardiovascular and ocular systems.

The common oral manifestations for which patients seek aesthetic correction from the dentist are constriction of the maxilla, crowded dentition, many missing teeth, and concomitant cross-bites, and this is when they are typically diagnosed. Furthermore, periodontal bone loss is linked to connective tissue problems as well.[1] Despite the lack of a proven link between severe periodontitis and Marfan syndrome, reports of alveolar bone loss and a high attachment level.

Another important factor is that these patients' heart pathology raises the risk of endocarditis after any dental procedure that could cause bleeding.[3] In our case also patient's sister had similar skeletal features. Though the patient had almost all the clinical signs of the Marfan syndrome and major criteria of Ghent score matched , the patient didn't show any of the cardiac features. The patient also showed low esteem regarding his aesthetic appearance.

The need for adequate at-home preventive measures for optimal oral hygiene, such as powered and customised toothbrushes, topical fluoride treatment, and antibacterial mouthwashes, must be emphasised to the patients.[3]

In our case, the orofacial characteristics helped to definitively diagnose MFS. It is widely acknowledged that these characteristics are more specific and that they should be used to rank individuals for proper referrals and in-depth examinations.

CONCLUSION :

Dentists need to be careful in identifying the oral symptoms of fibrillinopathies such as Marfan syndrome and other related craniofacial developmental diseases as unusual presentations as the management can require the use of multidisciplinary treatment focusing on the special needs of the patients.[1] The early diagnosis and medical management of the syndrome considerably increases patient's quality of life.

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