Acta Scientific Otolaryngology (ISSN: 2582-5550)

Volume 7 Issue 8 August 2025

Case Report

Primary Tooth Infraocclusion in Marfan Syndrome: Uncommon Presentation and Review of the Literature

Soumaya Kachti[1,2]*, Manel Chalbi[1,2] and Mohamed Ali Chemli[1,2]

[1]*Department of Pediatric Dentistry, La Rabta Hospital, Tunisia*

[2]*Faculty of Dental Medicine of Monastir, Tunisia*

***Corresponding Author:** Soumaya Kachti, Department of Pediatric Dentistry, La Rabta Hospital, Tunisia.

Received: June 19, 2025

Published: July 03, 2025

Abstract

Marfan syndrome is a hereditary connective tissue disorder with autosomal dominant transmission, known for its wide-ranging clinical manifestations across multiple organ systems. While the condition predominantly affects the skeletal, cardiovascular, and ocular systems, orofacial characteristics are also well described in the literature.

This article provides an in-depth review of the oral and dental features associated with Marfan syndrome. A comprehensive literature search covering the period from 1965 to 2024 was conducted using PubMed/Medline and Science Direct data bases. Keywords such as “Marfan syndrome”, “oral manifestations”, and “dental findings” were used in various combinations to retrieve relevant publications. Reference lists of selected articles were also screened manually to identify additional studies. Out of 126 articles initially identified, 21 met the inclusion criteria.

The review confirmed a variety of craniofacial and dental anomalies commonly linked to the syndrome. However, infraoccluded (submerged) teeth appear to be extremely rare, with only one case reported in 1965 involving a twin female patient. To date, this feature has not been widely acknowledged as part of the Marfan oral phenotype.

The present case report describes an 11-year-old child with Marfan syndrome presenting with infraocclusion of primary molars. This finding may expand the current understanding of orodental manifestations in Marfan syndrome. Management included extraction of the affected primary molars, followed by orthodontic referral for ongoing care.

Keywords: Marfan Syndrome; Dental Crowding

Introduction

Marfan syndrome (MFS) is an inherited disorder of connective tissue. A mutation of the fibrillin 1 gene (FBN1), localized in chromosome 15 (15q21) [1], leads to an increased level of Transforming growth factor beta (TGF-β) in tissues [2]. This gene encodes the elastic fibers, a major component of connective tissue. Hence Its mutation results in varied and combined symptoms of the heart, circulation, muscles, skeleton, eyes, and lungs [3].

This condition was named after a French pediatrician, Antoine Bernard-Jean Marfan, who first described its occurrence in 1896 [4].

The incidence of Marfan syndrome is approximately 1:5000 with no reported difference between gender, ethnic and geographic groups [5].

This syndrome may be asymptomatic. However, patients with this syndrome have some typical clinical manifestations including tall stature, long limbs, mitral valve prolapse, and aortic aneurysm development [6].

The orofacial symptoms of Marfan syndrome include a long and narrow face,enophtalmos, retrognathia, skeletal class II malocclusion, temporomandibular dysfunction [7].

Dental manifestations are mostly : highly arched palate with crowding of the teeth, posterior crossbite, malocclusion, and changes in the number of teeth [8].

The diagnosis of Marfan syndrome is based on a combination of the major and minor clinical features described in the 1986 Berlin classification system, which was revised by expert consensus to create the 1996 Ghent classification system [9]. Ocular, cardiovascular, and musculoskeletal abnormalities are considered the "classic triad" of MFS.

This article aims to describe a rare oral manifestation of this disease. We present a case report of a boy with infraoccluded deciduous teeth associated with MFS syndrome, along with a literature review.

Case Report

A.F, 11-year-old boy, Was adressed to the department of pediatric and preventive department, la rabta hospital tunis, by Physical medicine and functional rehabilitation department. The Chief complaint was « an inaesthetic anterior dental crowding ». The patient had been diagnosed with Marfan syndrome. Based on his medical history,The patient had ophtalmic problems (myopia), heart failure, and a mixed ventilatory deficit.

On general examination, the patient had tall stature, elongated extremities, Arachnodactyly of hands and feet. Dorso-lumbar scoliosis (Figure 1).

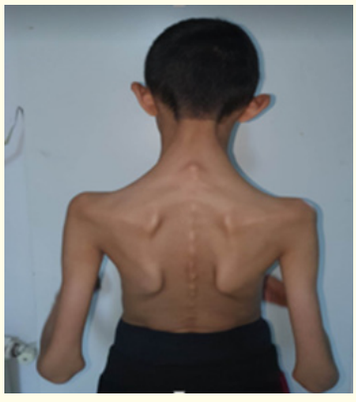

Figure 1: Dorso-lumbar scoliosis.

The patient had positive thumb (Steinberg sign: a flexed thumb clutched within a clenched palm protrudes beyond the ulnar border of that hand) evidence (Figure 2), and joint mobility.

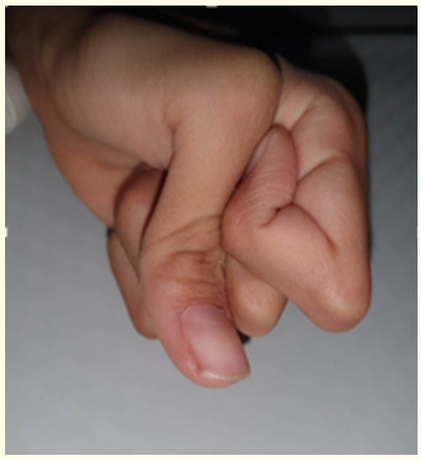

Figure 2: Steinberg sign.

The Craniofacial examination of the patient showed a convex profile, long face, and mandibular retrognathia (Figure 3).

Figure 3: Front and profile view.

Intraoral manifestations were mainly: High-arched palate, posterior crossbite on the left side, Maxillary and mandibular anterior crowding, Both the upper and lower midlines were aligned with the facial midline. Severly submerged deciduous teeth (84-85-74-75) (Figure 4,5).

Patient and her parents were informed about the treatment plan, and written and verbal consents were obtained.

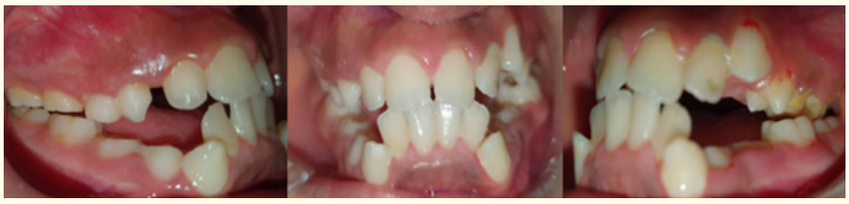

Figure 4: Intraoral view.

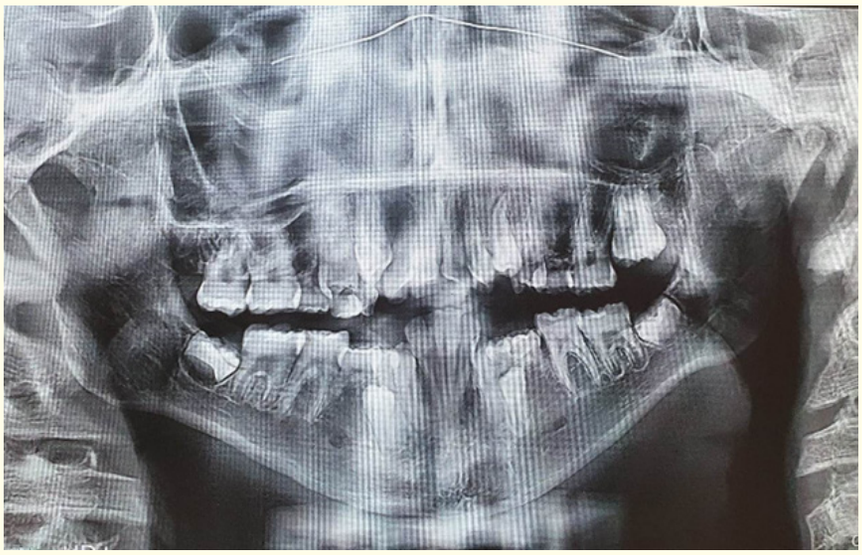

Figure 5: The Panoramic radiograph showed Agenesis of second permanent premolars (45-35-15-25).

The treatment plan was as follows: Extraction of infraoccluded first primary molars (74-84). The patient was addressed to an orthodontist. A regularly 6-month follow-ups are scheduled.

Discussion

Table 1 summarizes the key characteristics of the included studies.

A thorough search of literature from 1965 to 2024 was conducted using PubMed/Medline and ScienceDirect databases. The search strategy employed key terms such as «Marfan syndrome», «oral manifestations», and «dental manifestations» in various combinations. Citation lists from relevant references were examined, and additional reports were identified through hand searching. The search yielded 126 articles, of which 21 were included. Table 1 summarizes the key characteristics of the included studies.

Marfan syndrome is an autosomal dominant disorder that affects connective tissue. The disease is characterized by a wide range of clinical manifestations that affect different organs [31].

The syndrome primarily involves the malformations of skeletal, cardiovascular and ocular systems. The most common manifestations of the syndrome are disproportionately tall and slender structure, long arms, legs and fingers, pectus deformities and scoliosis, and often using eyeglasses because of severe myopia [32]. In the present case report, all these features were observed.

Authors	Year	Study Design	Gender (N)	Age (Years old)	Oro-facial manifestations
Sreekanth K., *et al*. [10]	2012	Literature review and Case report	Male	4,5	Supernumerary tooth, caries, maxillary arch was parabolic
Galletti., *et al*. [11]	2019	Systematic review and meta-analysis	-	-	Patients with Marfan's syndrome do not seem to have an aggravated oral health status.
Mariana C., *et al*. [12]	2010	Case report	Male	14	Very deep palatal, Class II molar relationship by Angle, DDM. generalized gingivitis caries, radicular cysts with radicular dilacerations, agenesia
Bostanci., *et al*. [13]	2017	Case report	Female (1)	7	Dolichocephaly, maxillary retrognathia, convex profile, high arched palate, midline shift, posterior crossbite, anterior teeth crowding, dental caries.
Khonsari., *et al*. [14]	2010	Case report	Female (1)	12	Broad forehead, highly arched palate, extreme dental crowding, supernumerary teeth, impacted teeth, crown and root malformations (narrowed pulp cavities loaded with a high number of pulpoliths).
Alam., *et al*. [15]	2022	Systematic review and meta-analysis	_	_	Marfan syndrome patients have narrow upper jaw, a high palate, mandibular retrognathism, crowding teeth. Also, they are more susceptible to periodontal inflammation.
Hanisch., *et al*. [16]	2018	Cross-sectional study	Male (17)	34.57 ± 17.58	Dysgnathia, malocclusion, high palate, crowding, hypodontia, mineralisation anomalies, microdontia, temporomandibular dysfunctions, and a poorer oral health-related quality of life in comparison to the general population.
			Female (34)	45.29 ± 11.90	
Suzuki., *et al*. [17]	2015	Case-control study	Male MG= 23 CG= 10	MG: 34.9 ± 2 CG: 32.4 ± 2.2	Marfan Syndrome subjects had lower remaining teeth, more frequent and more severe periodontitis when compared to non-affected ones.
			Female MG= 17 CG= 4	MG: 17 CG: 4	
Jain et Pandey [18]	2013	Case report	Female (1)	13	Dolicocephaly, mandibular retrognathia, malar hypoplasia, macrostomia, severe dental crowding, highly arched palate, anterior and posterior crossbite, gingivitis, carious tooth, and radicular malformations.
Shiga., *et al*. [19]	2017	Case report	Male (1)	11.5	Concave profile, long face, narrow dental arches, a posterior crossbite, deep overbite, high arched palate, two anterior supernumerary teeth, skeletal class III, and an inverted impacted maxillary central incisor.
Sinha., *et al*. [20]	2017	Case report	Female (1)	13	Dolichocephaly, long face, retrognathic profile, microdontia, enamel hypoplasia, deep palatal vault, dental crowding, excessive gingival display, increased overjet and overbite, delayed roots development on lower premolars.
Staufenbiel., *et al*. [21]	2013	Case-control study	Male MG: 21 CG: 14	MG: 40.20 ± 15.35 CG: 40.29 ± 13.94	MS patients did not exhibit a higher prevalence of periodontitis compared to the control group. But they are more prone to inflammation signs.
			Female MG: 30 CG: 17	MG: 40.20 ± 15.35 CG: 40.29 ± 13.94	

Tsang., *et al*. [22]	2013	Literature review and a case report	Female (1)	10	Maxillary and mandibular retrognathia, high arched palate, macroglossia, gingivitis, severe maxillary dental crowding, enamel opacities, pulpal calcifications, anterior and posterior crossbite, skeletal class III.
Bachani., *et al*. [23]	2016	Case report	Female	16	High arched palate DDM. caries ncreased overjet.
Sivasankari., *et al*. [24]	2017	Case report	Male (1)	22	Convex profile, high arch palate, anterior crowding teeth, poor periodontal status
Utreja., *et al*. [25]	2009	CASE SERIES	Female Male	12 13	Gingival inflammation and recession, protruding upper anterior teeth, generalized chronic gingivitis a supernumerary tooth II molar relationship with 13.5 mm overjet assymetric omega shaped maxillary arch anterior DDM
Nishikawa., *et al*. [26]	2013	Literature review	-	-	Retrognathia long face, and a highly arched palate, severe periodontitis
Cervino., *et al*. [27]	2020	Literature review	-	-	Dolichocephaly Susceptibility to periodontal diseases (Image 1) Oral calculus (Image 2 a, Image 2b) Root deformities Pulp anomalies Mandibular retrognathia Skeletal class II malocclusion Maxilla retrognathia Posterior crossbite Abnormal dentine formation Large positive overjet Hypermobility at the temporomandiular joints Malar hypoplasia Severe maxillary crowding Downslanting palpebral fissures Enophthalmos Maxillary constriction High/deep arched palate
Kamen., *et al*. [28]	1966	Case series	Female	8	Submerged second deciduous molars, denta caries, Caries, classe 2, narrow arches,high palatal vaults
De coster., *et al*. [29]	2002	Case control study	MG 23 CG 69	0-17	R caries. Local hypoplastic enamel spots were more frequent in Marfan syndrome and could be related to caries history of the deciduous dentition. Root deformity, abnormal pulp shape, and pulpal inclusions were a frequent finding in patients with Marfan syndrome.
Bauss., *et al*. [30]	2008		MG 21 10 male, 11 female) CG 100 47 male and 53 female	30	Pulp calcifications are frequent findings in subjects with Marfan syndrome.

Table 1

The updated Ghent nosology guidelines offer a framework that enhances clinicians' ability to accurately recognize Marfan syndrome cases [33].

The reported orofacial features in this case report were consistent to those reported in the literature, dolichocephaly, convex profile, high-arched palate, dental crowding which was anterior, and agenesis of mandibular second premolars were reported. However, this is the first case describing the presence of infraocclusion in primary teeth amongst patients affected with Marfan syndrome.

Infraocclusion is a term used to describe a tooth which has stopped its relative occlusal growth into the arch afer the period of active eruption; as a result, it becomes depressed below the occlusal plane [34].

Causes of infraocclusion identifed in the literature: Ankylosis, Other less common causes: Disturbed local metabolism Gaps in the periodontal membrane Local mechanical trauma Local Infection Chemical or thermal irritation Local failure of bone growth Abnormal tongue pressure Disturbance in interaction between normal resorption and hard tissue repair Radiation [35].

In the present case all lower deciduous molars were infraoccluded, the #75 and the #65 did not have successors, since the #45 and the #35 were agenesic. It is not an uncommon finding to have ankylosed primary molars with missing [36]. However this entity is rarely associated with MFS.

A late diagnosis of an early onset cases has a greater chance of complications. Treatment may include orthodontic uprighting of tipped adjacent teeth and subsequent restorative procedures. Studies advise orthodontic correction of tipped adjacent teeth to facilitate easier extraction if required and achieve favourable arch alignment [37]. Early diagnosis is the key to prevent these complications

As recommended in the litterature, The major goal of treatment of infraoccluded primary molars with successors is to allow the normal eruption of the successor [38]. Hence w. If exfoliation is not achieved within the accepted delay time of 6 months, extraction should follow The exrtraction of the #74 and #84 were preconised.

They also need an early and inter-orthodontic approach considering the many skeletal manifestations of the disease [36]. From here emerges the figure of the pediatric dentist to allow an early diagnosis of the diseases seen in the multiple oral manifestations. After extraction of the infraoccluded teeth the patient was referred to the orthodontic department.

Regarding the health of periodontal tissues, there are contrasting studies. De Costen's study states that the index of gingival inflammation is higher than that of the control group and for this reason also states arbitrarily that they are more susceptible to periodontal disease. This condition may be partly due to negligence on the part of such patients with regard to home oral hygiene maneuvers, and partly due to the etiopathogenesis of the disease itself, as well as the defect suffered by collagen and connective tissues [7].

Pediatric population still suffers today from a diagnostic delay of the MFS syndrome because the symptoms may not be evident, especially during growth. Consequently, pedodontists should be more involved in a multidisciplinary approach, to provide an early diagnosis of MFS at the paediatric ages.

Furthermore, dentists has an important role in terms of the risk of infective endocarditis that may occur during invasive dental treatments, which should not be overlooked [37].

Dental care for patients with Marfan syndrome is important to minimize the treatment need that could increase the risk of bacteraemia. Hence, periodic oral examination must be carried out.

Conclusion

Pedatric Dentists should be vigilant to recognize the oral manifestations of Marfan syndrome and other similar craniofacial developmental disorders. Timely diagnosis and early treatment help significantly improve the quality and longevity of these patients.

Knowledge about this rare syndrome's diagnosis, general health, and treatment modalities help pedodentists to offer more appropriate treatment for their patients.

Bibliography

1. Dean JC. "Management of Marfan syndrome". *Heart* 88.1 (2002): 97-103.

2. Benke K., *et al*. "The role of transforming growth factor-beta in Marfan syndrome". *Cardiology Journal* 20.3 (2013): 227-234.

3. McKusick VA. "Heritable disorders of connective tissue. III. The Marfan syndrome". *Journal of Chronic Diseases* 2.6 (1955): 609-644.

4. Utreja A and Evans CA. "Marfan syndrome: an orthodontic perspective".

5. Dietz HC., *et al*. "Marfan syndrome caused by a recurrent de novo missense mutation in the fibrillin gene". *Nature* 352.6333 (1991): 337-339.

6. Judge DP and Dietz HC. "Marfan's syndrome". *Lancet* 366.9501 (2005): 1965-1976.

7. De Coster PJ., *et al*. "Oral manifestations of patients with Marfan syndrome: A case-control study". *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology, and Endodontology* 93.5 (2005): 564-572.

8. Westling L., *et al*. "Craniofacial manifestations in the marfan syndrome: Palatal dimensions and a comparative cephalometric analysis". *Journal of Craniofacial Genetics and Developmental Biology* 18.4 (1991): 211-218.

9. De Paepe A., *et al*. "Revised dia gnostic criteria for the Marfan syndrome". *American Journal of Medical Genetics* 62.4 (1996): 417-426.

10. Mallineni SK., *et al*. "Concomitant occurrence of hypohyperdontia in a patient with Marfan syndrome: a review of the literature and report of a case". *Journal of Investigative and Clinical Dentistry* 3.4 (2012): 253-257.

11. Galletti C., *et al*. "Oral Health Status in Marfan Syndrome: A Systematic Review and Meta Analysis of 353 Cases". *International Journal of Environmental Research and Public Health* 19.9 (2022): 5048.

12. Mariana C., *et al*. "Case report on deep palate, Angle Class II molar relationship, generalized gingivitis, caries, radicular cysts with dilacerations, agenesis". (2010).

13. Bostanci H., *et al*. "Case report: dolichocephaly, maxillary retrognathia, convex profile, high-arched palate, midline shift, posterior crossbite, crowding, caries". (2017).

14. Khonsari RH., *et al*. "Extreme oral manifestations in a Marfan-type syndrome". *International Journal of Oral and Maxillofacial Surgery* 39.2 (2010).

15. Alam MK., *et al*. "Oral Health Status in Marfan Syndrome: A Systematic Review and Meta Analysis of 353 Cases". *International Journal of Environmental Research and Public Health* 19.9 (2022): 5048.

16. Hanisch M., *et al*. "Oral health and oral health-related quality of life in people with Marfan syndrome: a cross sectional study". *BMC Oral Health* 25 (2025): 925.

17. Suzuki S., *et al*. "Case control study comparing orofacial manifestations in Marfan (MG) vs control (CG) groups". (2015).

18. Jain M and Pandey RK. "Case report: dolichocephaly, mandibular retrognathia, malar hypoplasia, crowding, crossbite, gingivitis, caries, radicular malformations". (2013).

19. Shiga H., *et al*. "Case report: skeletal Class III, supernumerary teeth, narrow arches, high palate, impacted incisor". (2017).

20. Sinha S., *et al*. "Case report: dolichocephaly, long face, enamel hypoplasia, deep palate, crowding, delayed roots". (2017).

21. Staufenbiel I., *et al*. "Case control study: periodontal status and inflammation in Marfan syndrome". (2013).

22. Tsang AKL., *et al*. "Literature review and case report: retrognathia, macroglossia, crossbite". (2013).

23. Bachani S., *et al*. "Case report: high palate, DDM, caries, increased overjet". (2016).

24. Sivasankari T., *et al*. "Case report: convex profile, high-arched palate, periodontal issues". (2017).

25. Utreja A and Evans C. "Marfan syndrome—an orthodontic perspective". *Journal of Oral Science* 51.3 (2019).

26. Nishikawa T., *et al*. "Orofacial manifestations of congenital fibrillin deficiency". *Human Mutation* 19.1 (2004): 39-48.

27. Cervino G., *et al*. "Oral health status in Marfan Syndrome: review". *Archives of Oral Biology* (2020).

28. Kamen S., *et al*. "Case series 1966: submerged molars, caries, narrow arches". (1966).

29. De Coster PJ., *et al*. "Oral manifestations of patients with Marfan syndrome: a case control study". *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology, and Endodontology* 94.4 (2002): 420-426.

30. Bauss O., *et al*. "Prevalence of pulp calcifications in patients with Marfan syndrome". *Oral Surgery, Oral Medicine, Oral Pathology, Oral Radiology, and Endodontology* 106.1 (2008): 56-61.

31. Hirst AE and Gore I. "Marfan's syndrome: A review". *Progress in Cardiovascular Diseases* 16.2 (1973): 187198.

32. Rybczynski M., *et al*. "The spectrum of syndromes and manifestations in individuals screened for suspected Marfan syndrome". *American Journal of Medical Genetics* 146A.24 (2008): 3157-3166.

33. Loeys BL., *et al*. "The revised Ghent nosology for the Marfan Syndrome". *Journal of Medical Genetics* 47.7 (2010): 476-485.

34. Ekim S L., *et al*. "A treatment decisionmaking model for infraoccluded primary molars". *International Journal of Paediatric Dentistry* 11 (2001): 340-346.

35. Hua L., *et al*. "To extract or not to extract? Management of infraoccluded second primary molars without successors". *British Dental Journal* 227.2 (2019): 93-98.

36. Kula K., *et al*. "An occlusal and cephalometric study of children with ankylosis of primary molars". *Journal of Pedodontics* 8 (1984): 146-159.

37. De-La-Rosa-Gay C., *et al*. "Infraclusion of primary molars: Reports of cases". *ASDC. Journal of Dentistry for Children* 65 (1998): 47-51.

38. Nazif MM., *et al*. "The effects of primary molar ankylosis on root resorption and the development of permanent successors". *ASDC Journal of Dentistry for Children* 53 (1986): 115-118.