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(CASE REPORT)

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The dentist's role in diagnosing rare syndromes: Multidisciplinary management of dentoalveolar abscess in a 10-year-old child with Marfan syndrome

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Abstract

Marfan syndrome (MFS) is an autosomal dominant connective tissue disorder caused by mutations in FBN1 gene, characterized by significant cardiovascular pathology and multisystem fragility. A high level of caution is required in managing MFS in pediatric dentistry due to increased risk of infective endocarditis associated with valvular defects. This is a case report of a 10-year-old girl who presented with a submandibular space infection associated with the right mandibular first molar. Clinical evaluation revealed characteristic Marfanoid features, including a dolichocephalic facial profile, arachnodactyly, and the Steinberg sign. The tooth was extracted under strict antibiotic prophylaxis in adherence to American Heart Association guidelines. Following intervention, complete resolution of symptoms and uneventful healing were observed. This case emphasizes the pedodontist's pivotal role in the identification of orofacial manifestations and the early diagnosis of systemic syndromes. It further highlights the necessity of multidisciplinary coordination to ensure optimal outcomes for vulnerable pediatric patients.

Keywords: Marfan Syndrome; Infective Endocarditis; Pediatric Dentistry; Oral Manifestations.

1. Introduction

Marfan syndrome (MFS) is an autosomal dominant connective tissue disorder caused by mutations in the Fibrillin-1 (FBN1) gene on chromosome 15, with a prevalence of approximately 1 in 5,000. The condition is inherited in 75% of cases, while the remainder arises from sporadic mutations. Diagnosis follows the revised Ghent nosology (2010), underlining the need for early detection to render prophylactic medical and surgical interventions that have significantly extended life expectancy[1].

Dental management requires high clinical vigilance due to increased risk of infective endocarditis associated with underlying cardiovascular problems. Invasive procedures like extractions or periodontal surgery mandate strict adherence to antibiotic prophylaxis protocols to combat bacteremia. Local anesthetics should be selected judiciously, with felypressin preferred over epinephrine to mitigate the risk of cardiac acceleration[2]. To ensure patient safety and to avoid complex invasive dental treatment, preventive care, early intervention, and longitudinal monitoring are crucial.

2. Case report

A 10-year-old girl reported to the Department of Pediatric Dentistry with a primary complaint of a painful swelling on the lower right side of the face for three days. Despite a course of empirical antibiotic therapy initiated by a dental practitioner, the symptoms did not subside. Initial history-taking revealed parental reluctance to disclose the child's complete medical history. Upon comprehensive physical evaluation, classic clinical features of Marfan syndrome, including: Tall, slender stature with disproportionately long extremities (arachnodactyly) (Fig. 1), Steinberg

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sign/thumb sign (Fig. 2), Walker-Murdoch sign/wrist sign (Fig. 3), and joint hypermobility were noticed. Craniofacial evaluation showed a characteristic dolichocephalic (long, narrow) facial profile and malar hypoplasia. Unlike the typical presentation of mandibular retrognathism, prognathism was observed, which was confirmed by a lateral cephalogram (Fig. 4). A diffuse, firm, and tender swelling was noted over the right mandibular body, extending to the submandibular region.

Figure 1 Clinical Features

Figure 2 The Steinberg sign

Figure 3 Walker-Murdoch sign/wrist sign

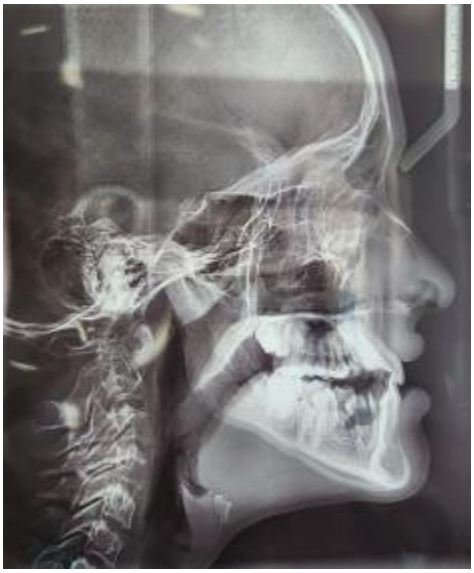

Figure 4 Lateral Cephalogram

Intraoral examination revealed a constricted, high-arched palate and severe dental crowding (Fig. 5). A deep carious lesion was noted in the permanent mandibular right first molar (46), which was exquisitely tender to percussion. An associated intraoral vestibular abscess was also noticed.

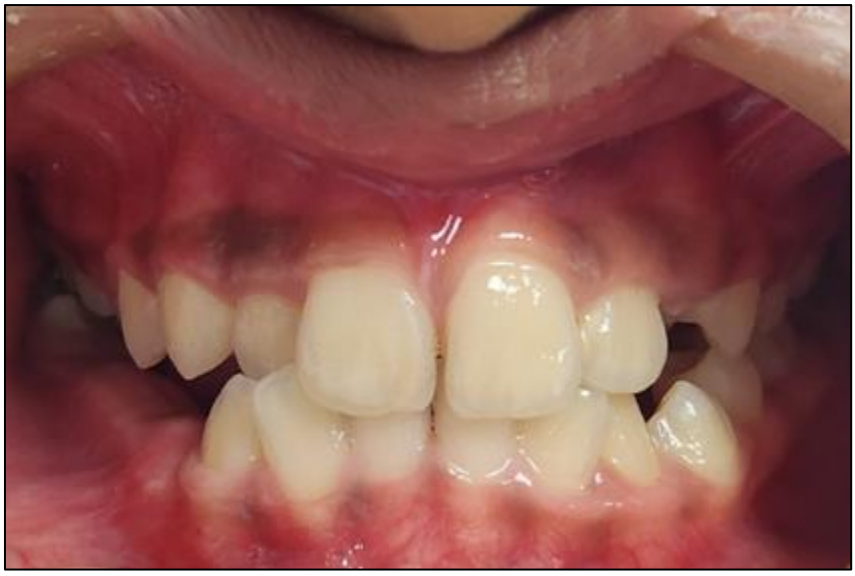

Figure 5 Intraoral Features

Marfan syndrome was diagnosed based on phenotypic findings and adherence to the Ghent nosology following a formal pediatric consultation. The primary clinical concern was the patient's susceptibility to Infective Endocarditis (IE) due to potential underlying valvular heart disease.

Management: After obtaining medical clearance by highlighting the systemic risk and taking informed consent, access opening of the tooth (46) was performed to facilitate drainage and symptomatic relief. Due to the frequent recurrence of the dental infection and the high cardiac risk status of the patient, extraction was planned to minimize the potential for systemic complications. The extraction was performed under strict antibiotic coverage according to AHA guidelines to mitigate IE risk. The extraoral swelling had completely resolved on the third postoperative day, and the healing was uneventful.

3. Discussion

The general features and oral manifestations of Marfan syndrome (MFS) must be understood as a downstream consequence of the underlying genetic pathophysiology. Fibrillin-1 is crucial for the formation of microfibrils, which are responsible for providing structural support to connective tissues and regulating the bioavailability of transforming growth factor-beta (TGF-β)[3]. The reduction or structural alteration of fibrillin-1 leads to hyperactivity of TGF-β signaling, resulting in weakened and disorganized connective tissues[1,2]. Aberrant TGF-β signaling serves as the primary driver for the Marfan phenotype, manifesting in the classic triad of skeletal overgrowth, ocular ectopia lentis, and cardiovascular abnormalities, specifically aortic root dilation and dissection[2,4].

Craniofacial manifestation of Marfan syndrome creates a distinct orthodontic challenge. The prevalence of high arched narrow palate is as high as about 69% in pediatric cohorts[4]. This often results in a transverse maxillary constriction leading to severe dental crowding. Consequently, posterior crossbites are more frequent in MFS patients, with one study noting a 2.5 to 7 times higher prevalence than in healthy controls[4]. The vertical growth pattern and ligamentous hyperlaxity affecting the temporomandibular joint (TMJ) often result in a Class II malocclusion and an anterior open bite. Additionally, intrinsic dental anomalies such as root deformities and pulp calcifications (pulp stones) may complicate orthodontic tooth movement[5,6].

The periodontal status of MFS patients remains a subject of debate in literature. Deficiency of Fibrillin-1, a major component of the oxytalan fibers, predisposes to disorganization and loss of integrity of PDL[7]. Based on histological studies, these microfibrillar defects can make the periodontium more susceptible to inflammatory breakdown[5]. However, there are conflicting data regarding the prevalence of periodontitis. Higher gingival inflammation indices in MFS patients compared to controls may not necessarily manifest more severe periodontal attachment loss. MFS is not an independent risk factor in the absence of other variables, as suggested by a recent meta-analysis wherein there was no statistically significant difference in pocket depth or bleeding on probing[8].In contrast, some studies link periodontal severity with the patient's cardiovascular status[7].

There is consensus that local plaque retentive factors associated with severe dental crowding in MFS patients could aggravate gingival inflammation, regardless of intrinsic tissue susceptibility[5,8]

MFS demands specific modification to dental management due to high systemic vulnerability, especially the risk of infective endocarditis (IE)[9]. Reducing the oral bacterial load is essential to minimize the risk of bacteremia-induced endocarditis[7]. While MFS itself is not a direct indication for antibiotic prophylaxis, the high prevalence of valvular pathology, especially the presence of prosthetic heart valves or grafts, mandates strict adherence to IE prophylaxis guidelines for invasive dental procedures[2].

Furthermore, local anesthesia should be cautiously administered. Epinephrine, the vasoconstrictor commonly used, should be discrete to avoid hemodynamic spikes that could increase wall stress on a dilated aorta or interact with beta-blocker medications commonly prescribed to these patients[2,13]. Surgical procedures like extractions pose higher risks for these patients. Anticoagulation therapy following valve replacement increases the potential for bleeding, while inherent tissue fragility can lead to delayed wound healing. Dural ectasia poses significant challenge in patient positioning for the procedure[1,2,3].

The dentist has a vital role in the multidisciplinary team. Craniofacial features such as the dolichocephaly, high-arched palate, and TMJ hypermobility are often evident early in life, before severe cardiac symptoms manifest[4,10]. These features, along with systemic signs like arachnodactyly, thumb (Steinberg Sign), and wrist signs (Walker-Murdoch Sign), should prompt immediate referral to the pediatrician for cardiovascular and genetic evaluation. The dental practitioners contribute significantly to the early diagnosis of Marfan Syndrome, facilitating timely medical intervention that can extend life expectancy[4,10].

4. Conclusion

The oral and craniofacial manifestations of Marfan syndrome (MFS) represent a critical intersection of systemic connective tissue pathology and oral health. The dental practitioner serves as a sentinel in the early diagnosis of MFS[10]. The responsibility of dentist's extends beyond the oral cavity to the multidisciplinary management[10].Proactive and effective oral health management does not merely preserve the dentition but serves as a fundamental shield for cardiovascular stability, remarkably enhancing the longevity and overall quality of life for individuals diagnosed with Marfan syndrome.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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