FACIAL AND OCCLUSAL GROWTH MANAGEMENT IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY – CASE REPORT

Manejo do crescimento facial e oclusal em pacientes com distrofia muscular de Duchenne – relato de caso

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Resumo

Pacientes com Distrofia Muscular de Duchenne (DMD) tendem a desenvolver mordida aberta anterior e crescimento mandibular excessivo (classe III) a partir dos 7 anos. *Objetivo*: Este estudo tem como objetivo apresentar uma abordagem terapêutica para esses pacientes, demonstrando, por meio de relato de caso clínico, uma estratégia de estabilização do desenvolvimento orofacial e da oclusão. *Relato de Caso*: K.V.J., do sexo masculino de 9 anos e 11 meses, com queixa inicial de cefaleia secundária à disfunção temporomandibular e apneia obstrutiva do sono (AOS) grave. Clinicamente apresentava mordida aberta lateral e tendência a mordida cruzada posterior. Paciente foi submetido à expansão rápida da maxila (ERM) para expansão maxilar, seguida do uso do aparelho miofuncional SN3 modificado por Vinha para controle da musculatura, especialmente da língua. Observou-se aumento do espaço das vias aéreas superiores, redução do índice de apneia-hipopneia (de 20,4 para 6,2), melhora das cefaleias e estabilização da oclusão, sem recorrência das alterações mencionadas anteriormente. *Considerações Finais*: O acompanhamento deste caso por mais de 8 anos demonstrou estabilidade facial e funcional, contrariando o padrão típico de deterioração associado à DMD. A intervenção precoce e integrada pode controlar o desenvolvimento orofacial e oclusal em pacientes com DMD, melhorando a estética, a função e a qualidade de vida.

Palavras-chave: Distrofia Muscular de Duchenne; Má oclusão; Ortopedia Funcional da Mandíbula; Apneia Obstrutiva do Sono; Aparelho Miofuncional

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Introduction

Muscular Dystrophy (MD) is a rare and progressive genetic disease characterized by the degeneration of skeletal, smooth, and cardiac muscles, leading to muscle weakness and loss of muscle mass[1]. It is caused by mutations in the gene that encodes the dystrophin protein, which is essential for the integrity of muscle cells. The absence or deficiency of this protein results in progressive muscle damage and loss of function[2]. As a progressive neuromuscular condition, it imposes multifaceted challenges on patients' health, and orofacial manifestations represent a critical and often underestimated component of this condition[3]. The most common type is Duchenne Muscular Dystrophy (DMD), inherited in an X-linked recessive pattern[4], which affects only male individuals with a prevalence of 1.04 per 10,000 live births[5].

DMD significantly affects craniofacial morphology and orofacial functional capacities[6]. In patients with DMD, a high prevalence of lateral and anterior open bites with decreased overjet, as well as posterior crossbites, has been identified[7,8]. Other studies have found a tendency toward excessive mandibular growth[9–11]. In general, most patients develop, from the age of 9 years onward, a condition of excessive mandibular growth and open bite, as can be observed in Figure 1.

Figure 1: Malocclusion features of a 16-year-old untreated patient with DMD.

It is known that these occlusal alterations directly impact the correct development of facial bones, combined with the muscle weakness present in these patients. In addition, the weakness of the dilator muscles of the upper airway may lead to respiratory disorders[12,13] related to sleep, such as obstructive sleep apnea (OSA)[14]. This condition can affect sleep quality, cause snoring, apnea, morning headaches, daytime hypersomnolence, as well as maxillary and mandibular bone alterations.

Specific symptomatic treatment and advances in cardiac and respiratory care have increased life expectancy to the extent that patients with DMD now reach 25–30 years of age[15]. As a consequence of increased life duration, regarding dentistry, difficulties in chewing and swallowing are demanding greater attention[16–18]. Few studies have encompassed the orofacial characteristics of patients with DMD, as it is a rare disease and the number of available patients is often limited, which can be even more reduced in longitudinal studies[19].

Due to these important facial and occlusal alterations, the earliest possible intervention becomes necessary, so that muscular changes do not negatively affect facial and occlusal development. Therefore, the objective of this study was to present the management adopted for treatment and to demonstrate a form of stabilization and maintenance of orofacial development, counteracting the natural tendency of patients with DMD, improving their oral functions, quality of life, and facial esthetics, through a clinical case.

Case Report

Ethical aspects

This study was approved by the Ethics and Research Committee of Hospital Moria under protocol number 67252822.0.0000.8054, in accordance with the requirements of the Brazilian National Health Council Resolution No. 466/2012.

Case Description

Patient K.V.J., male, diagnosed with DMD at 7 years of age, arrived at our service for malocclusion treatment at the age of 9 years and 11 months. He was referred by the Hospital das Clínicas of São Paulo, School of Medicine, University of São Paulo, with the chief complaint of recurrent and intermittent tension-type headache. In the dental clinical examination, the patient presented significant pain upon palpation of the temporal muscle, lateral open bite, and transverse maxillary deficiency. The mandible was tending toward Angle Class III (excessive anterior growth), with proclination of the lower incisors, as can be seen in Figure 2.

Figure 2 – Initial intraoral photographs of the patient at 9 years and 11 months of age.

In the lateral cephalometric radiograph, a narrowing of the airway was observed in the tongue base region. The PAS (posterior airway space) measurement, which evaluates the oropharynx, was only 2 mm (Figure 4). The patient presented with snoring during sleep, and polysomnography (PSG) confirmed that the child had severe obstructive sleep apnea (OSA), according to the American Thoracic Society. The patient had an apnea-hypopnea index (AHI) of 20.4, which is considered moderate for adults but severe for a child.

Proposed Intervention

Given the complex condition, rapid maxillary expansion (RME) was initially chosen, aiming to increase the transverse dimension of the dental arch and the nasal cavity. RME was performed in October 2016, when the patient was 9 years and 11 months old.

After removal of the fixed expander (Figure 3), an intraoral myofunctional appliance, called SN3 (Simões Network 3), modified by Vinha (Figure 4), was installed 10 months after the beginning of treatment.

Figure 3 – Frontal photograph after removal of the rapid maxillary expander.

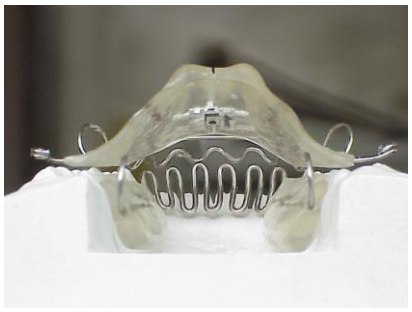

Figure 4 – SN3 modified by Vinha.

Initially, the appliance was used full time, and the patient maintained this regimen (active treatment) for 13 months. (Figure 5).

Figure 5: Frontal photograph after 13 months of full-time use of the functional appliance.

Figure 6 shows the photographs 6 months after nighttime use only, demonstrating case stability (March 2019).

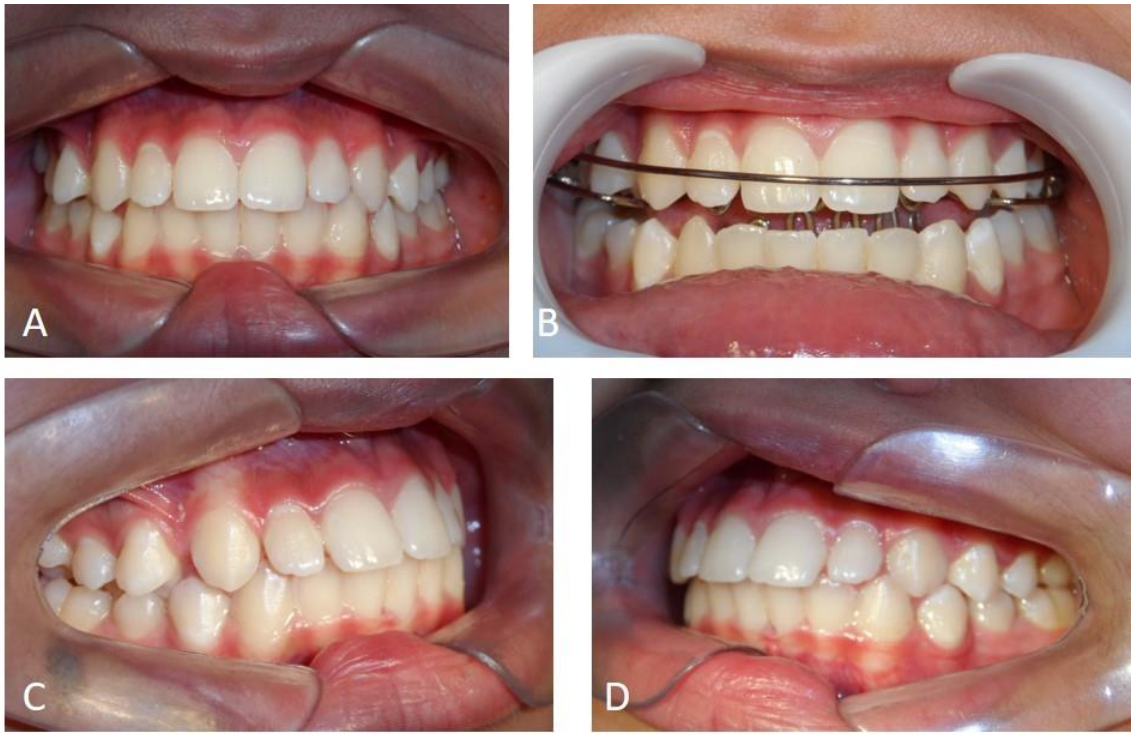

Figure 6: Frontal photograph without and with functional appliance (A and B), right and left lateral photographs (C and D).

After this period, the patient was discharged and instructed to use the appliance only during sleep, as retention, until March 2021, when the patient completely discontinued its use due to the pandemic.

After 15 months from the beginning of treatment, a new PSG and a new lateral cephalometric radiograph were performed to monitor OSA and mandibular growth.

Results

For the first intervention, which was carried out using the fixed appliance with occlusal coverage, a total maxillary expansion of 8.4 mm was achieved. In addition, there was a reported reduction in the frequency and intensity of headache episodes, and after the expansion was completed, the headaches practically ceased. In the second stage of intervention, with the aid of the SN3 modified by Vinha, occlusal stability and controlled growth were verified.

In the second PSG, a reduction of 69.60% in the apnea-hypopnea index (AHI) was observed (from 20.4 to 6.2). Other polysomnographic values also showed improvement, such as the arousal index and oxygen saturation. These values are shown in Table 1.

Table 1: Comparison of main polysomnographic values before and after RME, after 15 months.

	Initial	15 months
Sleep Efficiency	78.4%	81.9%
Arousal Index	19.9	6.6
Apnea Index	12.3	1.4
Hypopnea Index	8.0	4.8
AHI	20.4	6.2
Mean Saturation	93%	93%
Minimum Saturation	80%	84%
Time Below 90%	1.2 min	2.7 min

In the lateral cephalometric radiograph, an increase in PAS (distance between the base of the tongue and the posterior pharyngeal wall, along a line passing through point B and the Gonion) was observed (from 2 mm to 8 mm), as shown in Figure 7.

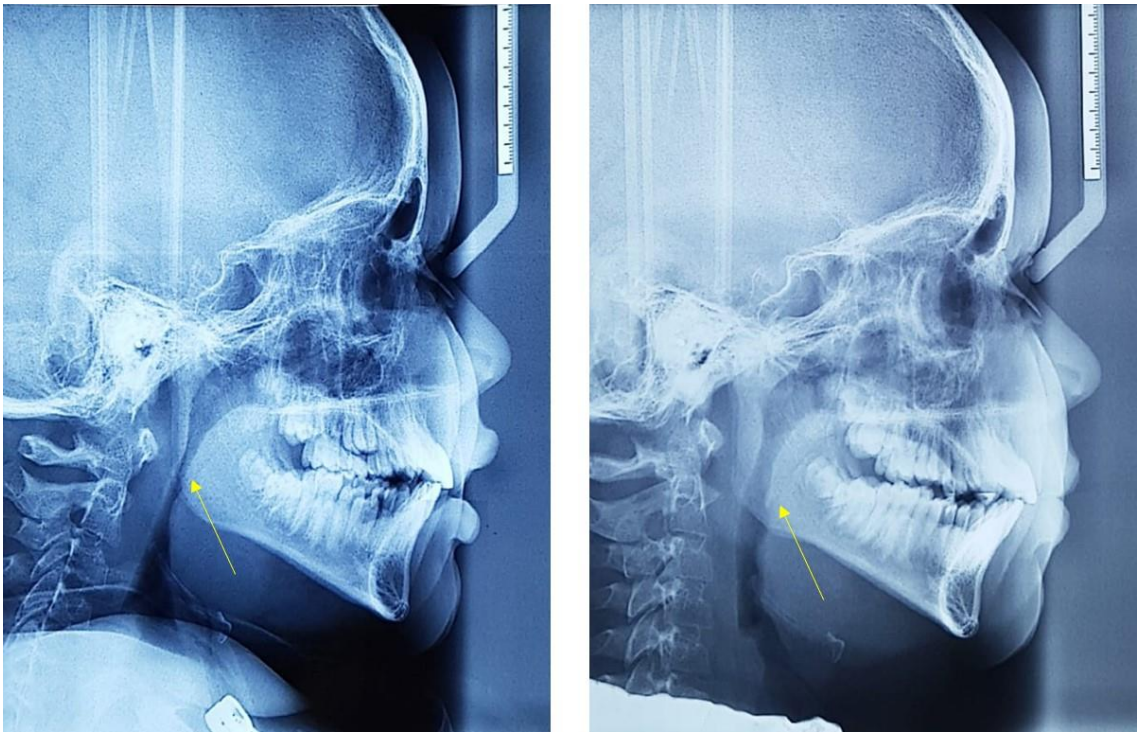

Figure 7 – Lateral cephalometric radiographs before and after treatment

In the images below (Figure 8), the case remains completely without the appliance and stable. Currently, the patient has been without any retention for 4 years and 4 months, with stable occlusion and facial development.

Figure 8 – Frontal, lateral, and upper and lower occlusal photographs.

Discussion

Patients with DMD present generalized muscle weakness that extends to the orofacial muscles, resulting in significant alterations in facial and occlusal development. These alterations notoriously compromise esthetics, masticatory function, phonation, and respiration[20]. The observation that life expectancy in patients with DMD has increased to 25–30 years, due to advances in cardiorespiratory care and symptomatic treatments[16–18], gives even greater urgency to the management of dental complications. Therefore, it is essential that dentists are prepared to manage these patients in collaboration with other medical specialties.

The occlusal alterations typical of DMD—including excessive mandibular growth, lateral and anterior open bite, and posterior crossbite—are the result of an imbalance between muscular forces. The early weakening of the muscles responsible for mouth closure, such as the masseter, in

contrast to the later atrophy of the orbicularis oris muscle, and the pseudo-hypertrophy of the tongue, create an environment conducive to these malocclusions[7,11,21], also leading to low masticatory development[20]. The described case, if it did not involve the systemic condition, would very likely have been relatively simple to manage with the aid of orthopedic and/or orthodontic therapies together with speech therapy. However, the remarkable aspect of this case was the control and stability of the occlusion despite a significant and persistent dysfunction: the progressive muscle weakness and degeneration present.

The intervention initiated with RME in the case of K.V.J. proved to be a fundamental strategy to address maxillary transverse deficiency. The results were clinically significant and multifaceted. RME not only promoted transverse dental arch expansion, but also expanded the nasal cavity, which translated into a notable improvement in polysomnographic (PSG) parameters and in the upper airway[22–25]. The reduction in the apnea-hypopnea index (AHI) from 20.4 to 6.2, although not fully normal, represents a substantial advance for a pediatric patient, moving him from a condition of severe OSA to a level considered moderate to mild. This improvement is of great importance, since untreated OSA can lead to cardiovascular risks and impaired cognitive performance, as described in the literature[26] and corroborated by the patient's own data (Table 1). Additionally, the increase in posterior airway space (PAS) from 2 mm to 8 mm, as visualized in the post-RME lateral cephalometric radiograph (Figure 8), supports the mechanism by which maxillary expansion contributed to improved airway patency.

The tongue in equilibrium with the perioral musculature is the best matrix for maxillary growth, provided that it is correctly positioned in the hard/soft palate region[27]. In patients with DMD, tongue positioning is altered[28–30]. Thus, for the second stage of treatment, the choice of the SN3 myofunctional appliance modified by Vinha after expansion was strategic, since this device is extremely effective for proper tongue posture, in addition to addressing the functional and muscular component underlying malocclusions in DMD. By promoting tongue repositioning and preventing its interposition between the dental arches, the SN3 modified by Vinha acts directly in stabilizing mandibular growth and preventing open bite, which are intrinsic tendencies in the progression of DMD. The maintenance of functional occlusion and pleasant facial esthetics, as evidenced by intraoral photographs of the patient 8 years and 9 months after the beginning of treatment (Figure 7), is testimony to the success of this myofunctional intervention in controlling orofacial development. The need for continuous initial use, followed by nighttime use for indefinite retention, underlines the chronic nature of management in patients with progressive conditions such as DMD.

The synergy between structural orthodontic intervention (RME) and the myofunctional approach (SN3 modified by Vinha) appears to be the key to the clinical success observed in this case. RME created the necessary space for respiratory improvement and facilitated transverse alignment, while the SN3 modified by Vinha addressed muscular dysfunctions and guided facial growth in a more harmonious and controlled manner. This combined approach is particularly relevant because DMD does not only affect bone structure, but primarily the muscular function that shapes these structures[2]. By intervening early, before changes become irreversibly established, it is possible to reprogram the orofacial environment, diverting it from the natural trajectory of deterioration expected in DMD.

It was also possible to observe, from the patient's report, remission of tension-type headaches after RME. Although not directly an outcome of DMD, this is an additional benefit that reflects improved sleep quality and reduced stress on the masticatory system, considering that headache was the patient's main referral complaint.

Although this study presents a single case, and not perfectly aligned to dental Class I, a clinical observation that at another time could be finalized with the aid of corrective dental appliances, it offers a promising model for orofacial management in patients with DMD. Even in the face of the progressive loss of muscle function inherent to the disease, it seems feasible to control occlusal and

facial changes, ensuring not only esthetics, but also the functionality of mastication, swallowing, and, critically, breathing. The preventive approach and continuous management demonstrated here may serve as a guide for future treatment protocols, emphasizing the importance of regular orofacial evaluation and interventions adapted to the individual progression of the disease.

The challenges for widespread implementation of this approach include the need for early diagnosis and intervention, patient and family adherence to long-term treatment, and coordination among a multidisciplinary team that includes neurologists, pulmonologists, orthodontists, and speech therapists. Furthermore, although the results are encouraging, the nature of a case study limits the generalization of findings. It is imperative that future studies, with larger cohorts and longitudinal designs, be conducted to validate this methodology in a broader population of patients with DMD.

In summary, this report reinforces the premise that orthodontic and myofunctional intervention, when applied in a timely and integrated manner, can offer significant improvement in the quality of life of patients with DMD, transforming a prognosis of orofacial deterioration into a scenario of functional and esthetic control and stabilization.

Conclusions

Based on the evidence presented in this case study, follow-up over more than 8 years demonstrated facial and functional stability, contradicting the typical pattern of deterioration associated with DMD. Early and integrated intervention can control orofacial and occlusal development in patients with DMD, improving aesthetics, function, and quality of life.

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Abstract

Patients with Duchenne Muscular Dystrophy (DMD) tend to develop anterior open bite and excessive mandibular growth (Class III pattern) from around 7 years of age. ***Objective:****. This study aims to present a therapeutic approach for these patients and to demonstrate, through a clinical case report, a strategy for stabilizing orofacial development and occlusion.* ***Case Report:*** *A 9-year-11-month-old male presented with headache associated with temporomandibular disorder and severe obstructive sleep apnea (OSA). Clinical examination showed lateral open bite and a tendency toward posterior crossbite. The patient was treated with rapid maxillary expansion (RME), followed by a Vinha-modified SN3 myofunctional appliance for muscle control, especially of the tongue. Outcomes included increased upper airway space, reduction in apnea-hypopnea index (from 20.4 to 6.2), improvement in headaches, and occlusal stabilization without recurrence.* ***Final Considerations:*** *After more than 8 years of follow-up, facial and functional stability was observed, differing from the typical progression of DMD. Early and integrated intervention may contribute to controlling orofacial and occlusal development, improving function and quality of life.*

Keywords: Duchenne Muscular Dystrophy; Malocclusion; Functional Orthopedics of the Mandible; Obstructive Sleep Apnea; Myofunctional Appliance

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