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CASE PRESENTATION

Alteration of the palate in myotonic dystrophy type 1. A case report

Alteración del paladar en la Distrofia Miotónica tipo 1. A propósito de un caso

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ABSTRACT

Introduction: myotonic dystrophy type 1 is a chronic, hereditary disease. It typically manifests during the third or fourth decade of life; however, phenotypic variability is broad, and it may be associated with palatal alterations unrelated to hypertrophy of the maxillary tuberosities.
Objective: to present a case of a patient with myotonic dystrophy type 1 and palatal alterations—specifically, unilateral hypertrophy of the maxillary tuberosity.
Case presentation: a 65-year-old female patient with third-degree atrioventricular block treated with a pacemaker and a medical history of hypertension, hip fracture, and cataract. Clinical signs of myotonic dystrophy type 1 are observed, including facial, palatal, and muscular abnormalities: generalized muscle weakness, myotonic phenomenon, and difficulty walking. The skin shows hyperhidrosis and hypopigmented macules. There is an increase in volume in the upper left maxilla with ulceration; diagnosis: hypertrophy of the left maxillary tuberosity, torus palatinus, and partial anodontia.
Conclusions: this case report highlights an additional palatal alteration to consider in patients with myotonic dystrophy type 1. These individuals face multiple risk factors—both biological and social—that may influence the association of both conditions.

Keywords: Myotonic Dystrophy; Maxilla; Gingival Overgrowth; Gingival Hypertrophy.

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RESUMEN

Introducción: la distrofia miotónica tipo 1 es una enfermedad crónica y hereditaria. Generalmente se presenta entre la tercera o cuarta década de la vida, sin embargo, existe amplia variabilidad fenotípica, puede tener asociado alteraciones del paladar no relacionado con la hipertrofia de las tuberosidades maxilares.
Objetivo: presentar a una paciente con Distrofia Miotónica tipo 1 con alteraciones del paladar: hipertrofia unilateral de la tuberosidad del maxilar.
Presentación de caso: paciente de 65 años con bloqueo auriculoventricular grado III tratado con marcapasos y antecedentes de hipertensión, fractura de cadera y catarata. Presenta signos de distrofia miotónica tipo 1 con alteraciones faciales, palatinas y musculares, incluyendo debilidad generalizada, fenómeno miotónico y dificultad para caminar. La piel muestra hiperhidrosis y máculas hipopigmentadas. Se detecta aumento de volumen maxilar izquierdo con ulceración; diagnóstico: hipertrofia de tuberosidad, torus palatino y anodoncia parcial.
Conclusiones: El reporte de este caso hace evidencias de otra alteración del paladar necesaria a tener en cuenta en los pacientes con Distrofia Miotónica tipo 1, en estas personas hay muchos factores de riesgo no solo biológicos sino también sociales que pueden condicionar la asociación de ambos trastornos.

Palabras clave: Distrofia Miotónica; Maxilar; Sobrecrecimiento Gingival; Hipertrofia Gingival.

INTRODUCTION

Myotonic dystrophy type 1 (DM1) is included in the group of neuromuscular diseases and its symptoms and signs are secondary to the involvement of a component of the motor unit: the muscle.[1] It is a chronic and hereditary disease. It generally presents between the third or fourth decade of life, however, there is wide phenotypic variability which is why it is considered a multisystem disease. According to the age of onset and the severity of the disease, some clinical forms can be delimited, among these are the congenital and the classic.[2]

The adult clinical form presents between the ages of 20 and 40 years and is characterized by muscle weakness and atrophy, as well as myotonia. An evident and early characteristic is the typical physical appearance (myopathic facie) caused by weakness and atrophy of the facial muscles, eyelid ptosis that gives the false impression of tiredness, sadness or emotional apathy. This is complemented by baldness and atrophy of the temporal muscle that determines an incomplete mouth closure. A high-arched palate and other stomatological problems conditioned by the decrease in oral hygiene are also frequent.[3]

On the other hand, fibrous hyperplasia (FH) of the maxillary tuberosities is a localized manifestation of a clinical entity known as gingival fibromatosis (GF). GF is a diffuse hyperplasia of the maxillomandibular gingiva. The thickening of the gingival tissues can be localized or generalized. Localized forms are usually located on the maxillary tuberosities or on the buccal or lingual surfaces of the mandibular molars. It can occur in isolation or in association with numerous syndromes.[4]

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This rare entity has received different names in the literature, mainly referring to the generalized forms. It is known as: gingivomatosis, elephantiasis, common elephantiasis, diffuse fibroma, idiopathic fibromatosis, hereditary gingival fibromatosis, congenital common fibromatosis or hereditary gingival hyperplasia.[5,6] Among the causes are systemic diseases, antiepileptic drugs (phenytoin), immunosuppressants (cyclosporine), antihypertensives (nifedipine, diltiazem and verapamil), inflammation and poor management of adequate hygiene techniques at home by the patient or their caregivers in the case of dependent patients. The prognosis of gingival fibromatosis will depend on oral prophylaxis at home.[7]

Complications associated with gingival fibromatosis include difficulty chewing, speech problems, occasional tooth displacement, aesthetic defects, and psychological difficulties. It also causes prolonged tooth retention in children, which makes oral hygiene and plaque control difficult. With this in mind, the present study aims to present the case of a patient with Myotonic Dystrophy type 1 with palatal abnormalities: unilateral maxillary tuberosity hypertrophy.

CLINICAL CASE REPORT

This is a 65-year-old patient who has been diagnosed with third-degree atrioventricular block for 15 years, for which she has a pacemaker. She has a history of high blood pressure, for which she has been treated with several antihypertensives. She has undergone surgery for a hip fracture and a cataract in her right eye.

Dysmorphological examination reveals signs of myotonic dystrophy type 1 with palatal alterations, which are the following: weakness of the facial muscles, myopathic facies, facial asymmetry, broad forehead, periorbital hyperpigmentation, bilateral eyelid ptosis, hypoplasia of the nasal wings, prominent columella, poor dentin quality, palatine torus and increased volume in the upper region of the left maxilla with an area of ulceration (Fig. 1). In addition, the presence of generalized muscle weakness, myotonic phenomenon in both hands, difficulty walking. Thin, dry skin, hyperhidrosis of both hands with hypopigmented macules on the trunk and upper limbs.

Fig. 1 Increased volume in the upper region of the left maxilla with an area of ulceration and palatine torus.

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Given this finding in the left upper jaw, a consultation was made with a maxillofacial specialist who, after a physical examination, concluded a diagnosis of hypertrophy of the left maxillary tuberosity with an ulcerated area and palatine torus, in addition to partial anodontia.

DISCUSSION

The 2022 fifth edition of the WHO Classification of Tumours of the Head and Neck has added segmental odontomaxillary dysplasia (DOMS) to the group of fibro-osseous lesions. This classification includes disorders such as non-progressive or slowly progressive overgrowth or undergrowth of soft and/or bony tissues (typically affecting the posterior maxilla and leading to facial asymmetry), dental anomalies (missing teeth or abnormal dentition), and gingival tissue abnormalities.[8]

Fibrous hypertrophy of the maxillary tuberosities is a localized manifestation of a clinical entity known as gingival fibromatosis and may be an isolated entity or part of a syndrome or chromosomal abnormality.[2,9] The association of maxillary tuberosity hypertrophy with myotonic dystrophy type 1 is not recorded in the reviewed literature, but it is not difficult to relate both conditions. In DM1, progressive osteomyoarticular involvement generates mixed disabilities and cognitive deterioration sets in, which often leads to lack of oral health care. These social aspects, together with drug ingestion for other associated chronic diseases such as hypertension, constitute risk factors that predispose to palate disorders.

However, it is necessary to make a differential diagnosis of maxillary tuberosity hypertrophy with the hereditary form, which has a genetic origin and is autosomal dominant. It is rare and is characterized by a benign, slow-progressing, non-hemorrhagic fibrous enlargement that affects the maxillary and mandibular gingiva. In the patient in this case, there is no family history of these palate disorders.[8] Other syndromes to consider are: Zimmermann-Laband syndrome, Jones syndrome, Ramón syndrome, juvenile hyaline fibromatosis, and systemic infantile hyalinosis.[9]

It generally exists as an isolated anomaly; in the reviewed literature no association with DM1 is found. In myotonic dystrophy the reported craniofacial alterations are secondary to defects in the osteomyoarticular system and the most frequent alterations are the ogival palate, alterations in the dental arrangement, disorders in the quality of the dentin and secondarily deficiencies in oral hygiene.[10] In the research published by Licourt Otero and collaborators,[11] no other patients with DM1 were found who presented hypertrophy of the maxillary tuberosity.

Maxillary tuberosity hypertrophy can interfere with mastication and lip closure, causing difficulty speaking, as is the case with this patient. The pathogenesis is not well understood. Research suggests that the pathogenesis is limited to fibroblasts in the gingiva. Decreased apoptosis, along with increased proliferative activity in fibroblasts, may contribute to fibrotic overgrowth.[11] According to some studies, the correlation between the number of fibroblasts and collagen remains controversial. Impaired collagen production and degradation may also contribute to the disease.

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Molecular studies show abnormal expression of some molecules related to extracellular matrix metabolism, for example, transforming growth factor-β, which leads to increased deposition in the extracellular matrix that contributes to the pathogenesis. With the development of molecular genetics, maxillary tuberosity hypertrophy has been linked to chromosome 2p21-p22 and 5q13-q22. A maturation in the Sevenless 1 (SOS-1) gene has been identified, which may be responsible for the isolated form of hypertrophy.[12]

The genetic mechanism is not fully understood; therefore, most research has credited this condition with hereditary factors.[9,12] The enlargement can also be drug-induced, which may explain the presence of this alteration in the patient, or caused by local factors such as plaque and calculus, and associated with systemic, neoplastic, or idiopathic factors. Furthermore, it has a progressive growth that patients report as increasing over time, as this patient reports.

CONCLUSIONS

This case report highlights another palatal abnormality that should be considered in patients with myotonic dystrophy type 1. These individuals have many risk factors, not only biological but also social, that can lead to the association of both disorders. There is contradiction in the literature regarding the cellular and molecular mechanisms that lead to maxillary tuberosity hypertrophy. It generally presents in isolation, but periodic physical examinations are essential in patients with type 1 DM to diagnose early rare palatal abnormalities that may be associated with the disease.

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