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ORIGINAL ARTICLE

Orofacial Disorders in Myotonic Dystrophy Type 1

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ABSTRACT

Introduction: myotonic dystrophy type 1 is a genetic disease in which there are orofacial disorders with variable expression between the different clinical forms of the disease.
Aim: describe orofacial disorders in myotonic dystrophy type 1.
Methods: a descriptive, cross-sectional and retrospective study was carried out in the provincial department of Medical Genetics, of the "Pepe Portilla" Pediatric Hospital in the province of Pinar del Río, from January 2019 to December 2023. We worked directly with a population of 97 patients, without sampling.The documentary review and the physical examination allowed obtaining information that gave rise to the variables analyzed, using descriptive statistical methods..
Results: a high-arched palate was common in all clinical forms; however, upper lip eversion was less common. Incomplete mouth closure predominated in the most severe clinical forms of the disease, while the myopathic face was more common in all clinical forms, except in the late clinical form, where sunken cheeks predominated. Frontal baldness was revealed in patients with the infantile clinical form..
Conclusions: orofacial disorders in DM1 are common and cause not only aesthetic alterations but also disruptions in essential processes such as chewing and swallowing. Early identification and treatment are essential to prevent complications affecting various organs and systems.

Keywords: Mouth Abnormalities; Myotonic Dystrophy; Facial Expression.

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INTRODUCTION

DM1 is a genetic disorder whose mutation has its locus on chromosome 19q13.3. The mutation of the CTG (cytosine, thymine, guanine) triplet of the DMPK gene encodes the protein kinase enzyme whose function is related to muscle contraction and its elevation suggests the destruction of muscle fibers.[1,2]

In patients with myotonic dystrophy type 1 (DM1), the expansion of the CTG trinucleotide can vary from 50 to several thousand repetitions, and this variability is directly correlated with the severity of clinical symptoms: the greater the number of repetitions, the more severe the manifestation tends to be. According to this relationship, a clinical classification is established that includes the congenital form, with between 1000 and 2000 repetitions; the classic form, which ranges between 50 and 1000 repetitions and is associated with the infantile, juvenile and adult variants; and the late form, in which the expansions range between 50 and 100 repetitions. In addition, some asymptomatic individuals may carry the mutated allele with between 38 and 49 repetitions, which does not generate evident phenotypic manifestations but does imply an increased risk of transmission to offspring.[3,4]

Muscular dystrophies have in common an etiology that is due to the absence, reduction or dysfunction of proteins essential for the structural and functional stability of skeletal muscle fibers, which leads to destruction and muscle weakness in a progressive manner. Myotonic dystrophy combines this characteristic plus myotonia, therefore it is included in the group of neuromuscular diseases and its symptoms and signs are secondary to muscle involvement.[5] The disorder has a wide phenotypic variability so it is considered a multisystemic disease.[6]

Muscle weakness and atrophy are progressively established in the facial muscles, which contributes to the facial dysmorphisms observed in these patients.[7] The mouth is usually triangular in shape, with the upper lip retracted and the lower lip rotated outwards, and often remains open. In the literature, it is reported that the craniofacial development of many people with DM1 is characterized by more vertical cranial growth, narrower maxillary arches and a deeper palate than in healthy controls, and that malocclusion, especially frontal open bite and crossbite, is common. Taking into account the above, the present research is developed, which aimed to describe the orofacial disorders in patients with myotonic dystrophy type 1.

METHODS

A descriptive, cross-sectional and retrospective study was conducted in the provincial department of Medical Genetics of the "Pepe Portilla" Pediatric Hospital in the province of Pinar del Río, Cuba, from January 2019 to December 2023.

The study universe consisted of all people, of any sex and age with DM1, who resided in the province of Pinar del Río. The study population included 97 affected individuals. When applying the inclusion criteria (informed consent for participation in the study and inclusion in the registry of genetic diseases of the province of Pinar del Río), all patients gave their consent for the research, which allowed to form a sample with the 97 cases.

Patients were classified according to the clinical forms of the disease that take into account the age of onset of clinical manifestations: congenital (less than one year of age), infantile (1-10 years), juvenile (11-20), adult (21-40) and late (41 years and older). A physical examination was performed to look for orofacial alterations, disorders of the oral region included: ogival

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palate, incomplete mouth closure, altered dental arrangement, eversion of the upper lip, eversion of the lower lip, thick lower lip, upper lip in the shape of an inverted V, defective oral dental hygiene. As part of the facial alterations, the following were sought: inexpressive and myopathic face, palpebral ptosis, sunken cheeks, large and detached ears, as well as frontal baldness. There was a group of facial dysmorphisms classified as others, which included dolichocephaly, asymmetry, atrophy of the infraorbital muscles with hyperpigmentation and prominent columella.

Statistical Processing

The collected clinical data were entered and analyzed in the statistical package SPSS (Statistical Package for the Social Science) version 22. Qualitative variables were summarized with absolute frequencies and percentages, represented by tables and graphs.

Ethical Aspects

For this research, the approval of the Ethics Committee for Scientific Research of the Pepe Portilla Pediatric Hospital in the Province of Pinar del Río was obtained. In the process of taking informed consent, general information regarding the research was disclosed. The agreements approved by the Nuremberg Code of 1947 and the Declaration of Helsinki of the World Medical Association were respected, in these are embodied the ethical principles that must be taken into account for the realization of medical research with human beings.[8]

RESULTS

Table 1 shows the frequency of DM1 and craniofacial dysmorphism, according to clinical forms. There was a predominance of the congenital and adult clinical form. In all patients with onset of the disease below 20 years of age, craniofacial dysmorphisms were present.

Table 1. Frequency of DM1 and craniofacial dysmorphism, according to clinical forms.

Clinical forms (age in years)	Cases with DM1		Percentage of cases with craniofacial dysmorphism
	No.	**%**	
Congenital (< 1)	15	15,5	100
Infantile (1-10)	7	7,2	100
Juvenile (11-20)	12	12,4	100
Adult (21-40)	33	34	87,9
Late (41-84)	30	30,9	83,3

In Figure 1, the dysmorphisms in the oral region are shown according to clinical forms of the disease. For better representation, each clinical form was isolated in a graph (1A-Congenital, 1B-Infantile, 1C-Juvenile, 1D-Adult, 1E- Late).

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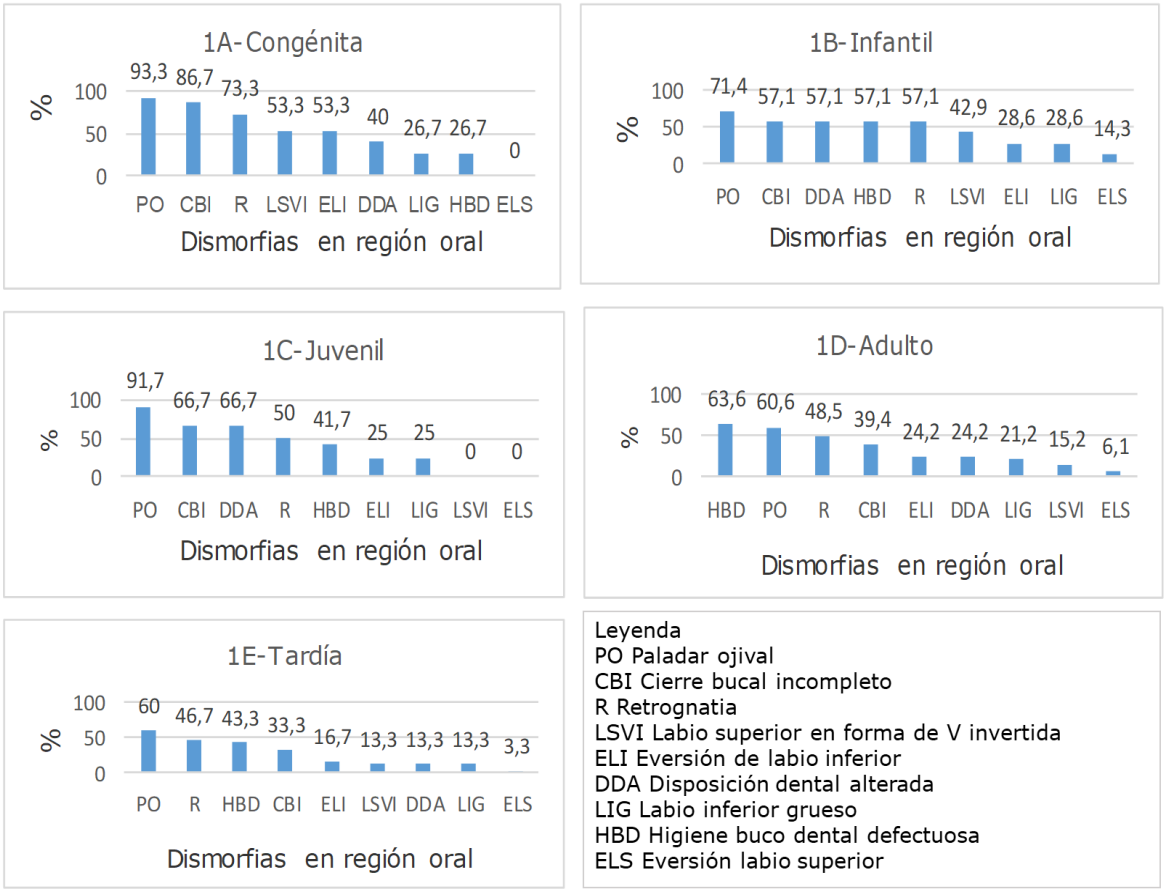

Fig. 1 Dysmorphisms in the oral region according to clinical forms (1A-Congenital, 1B-Infantile, 1C-Juvenile, 1D-Adult, 1E- Late)

The dysmorphisms of the facial region are described in Table 2. Myopathic face was more frequent in all clinical forms, except in the late form in which sunken cheeks predominated. Frontal baldness was revealed in patients with the infantile clinical form. There was a group of craniofacial dysmorphisms classified as others, which included dolichocephaly, asymmetry, atrophy of the infraorbital muscles with hyperpigmentation and prominent columella that had a high frequency in all clinical forms of the disorder. 100 % of patients with DM1 presented alteration of facial expression.

Table 2. Dysmorphisms in the facial region, according to clinical forms of DM1.

Facial dysmorphisms	Congenital		Infantile		Juvenile		Adult		Late	
	No.	%	No.	%	No.	%	No.	%	No.	%
Inexpressive face	6	40	5	71,4	6	50	23	69,7	18	60
Myopathic face	8	53,3	5	71,4	10	83,3	26	78,8	14	46,7
Sunken cheeks	7	46,7	2	28,6	4	33,3	23	69,7	23	76,7
Large ears	2	13,3	1	14,3	1	8,3	4	12,1	9	30
Detached ears	4	26,7	4	57,1	4	33,3	5	15,2	10	33,3
Frontal baldness	7	46,7	6	85,7	9	75	25	75,8	21	70
Others	15	100	6	85,7	10	83,3	27	81,8	21	70

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Rev Ciencias Médicas. 2025; 29(2025): e6865

DISCUSSION

In DM1, the clinical form that is best characterized is that of the adult with a frequency of 34 %, which coincides with the present study. Sánchez-Tejerina D et al,[9] declare a frequency of 66,4 %. The congenital clinical form has a frequency ranging from 4,5 % to 30 %,[1,2] which agrees with what was found in Pinar del Río.

For the infantile clinical form, its frequency is reported between 9,5 and 14,8 %.[9] The onset in the juvenile stage ranges between 30 and 36,5 % of patients. On the other hand, from 9,5 % to 16 % of people debut with the late form of the disorder.[2] When comparing the frequencies with the present study, it was considered lower for the infantile and juvenile forms, however in the late form the frequency of the disorder was higher, which can be explained because in the late form the clinical manifestations have a slight expression that leads to greater survival of the patient and also covered a wide range of ages (more than 40 years) that includes a greater number of people and it is likely that there is a greater number of patients with the disease.

Mira Escolano MP et al,[10] indicate for the region of Murcia in Spain, a frequency of 39,8 % for the adult clinical form, followed by the juvenile (18,8 %) and the late (16,7 %). The least frequent form is the congenital (2,9 %). With respect to the craniofacial dysmorphisms that are observed in people with DM1, it is necessary to highlight the muscle weakness and atrophy that are progressively established in the facial muscles.(7) In the literature that was reviewed, there are few studies that detail the craniofacial dysmorphisms by clinical forms.

In the article published by De Antonio et al,[11] they find facial dysmorphisms more frequently in the congenital form (82,1 %) and in the infantile form (61,6 %). Patients with DM1 present a greater vertical facial growth compared to the normal population.[12] The involvement of the facial musculature could be the cause of the mandibular rotation in a clockwise direction, either by gravity or by the absence of suprahyoid muscle support. This mandibular movement affects the position of the tongue and the posture of the head. The tongue, when positioned lower in the oral cavity, cannot counteract the forces created by the altered facial musculature.[13]

These new conditions could affect the transverse position of the teeth, reducing the palatal width and causing posterior crossbite.[14] In combination with the decrease in occlusal forces, the new postural position of the mandible could contribute to the overeruption of the maxillary posterior teeth. In this case, the palatal depth increases due to overeruption. The lower jaw can rotate even further in a clockwise direction, which contributes to the increase in the angle between the mandibular plane and the palatal plane.[15]

Evlice et al,[16] evaluated patients between 18 and 24 years of age, who have a high frequency of crossbite (42,4 %). Changes in dentofacial characteristics in the disease affect the oral function and oral health of patients. The facial and occlusal characteristics of patients with DM1 resemble the "adenoid face", where neuromuscular adaptation contributes to the backward mandibular rotation and the change in head posture to facilitate oral breathing. In this disorder, difficulties in chewing and swallowing are reported, patients with DM1 need 2,5 times more time to chew and have masticatory cycles 2,5 times longer than patients without the disease. In combination with the position of the mandible and tongue, this could precipitate the aspiration of food into the bronchial tree.[17,18]

ISSN: 1561-3194 **RNPS:** 1877
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Impaired facial expression due to weak and hypotonic facial muscles is a characteristic feature of the disease. Chawla T et al,[19] describe the myopathic face in more than 60% of cases, in all clinical forms, but it is more frequent in those patients with the adult clinical form; frontal baldness is another trait that exhibits a frequency greater than 70% in patients with the adult and late clinical form.

Palpebral ptosis is a dysmorphic sign that affects around 60 % of patients, it is produced by weakness of the levator muscle of the eyelid, it is usually constant in this disease and has a progressive course.[20] In the present investigation, this sign also showed a high frequency for all clinical forms, although higher in the adult.

CONCLUSIONS

Orofacial disorders in DM1 are frequent and produce not only aesthetic alterations but also disorders in essential processes such as chewing and swallowing. Identification and treatment from an early age are a fundamental pillar to avoid complications in different organs and systems.

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Conflict of Interests
The athors declare that there is no conflict of interest.

Authorship Contribution
DJC: Conceptualization, Research, Methodology, Project Administration, Original Draft Writing, Review and Editing Writing.
DLO: Conceptualization, Research, Methodology, Project Administration, Original Draft Writing, Review and Editing Writing.
ZTT: Research, Project Administration, Original Draft Writing.
LEMF: Research, Original Draft Writing.

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