Myotonic dystrophy and craniofacial morphology: clinical and instrumental study

M. PORTELLI, G. MATARESE, A. MILITI, R. NUCERA, G. TRIOLO, G. CORDASCO

Abstract. Aim The aim of this study is to assess if, and to what extent, myotonic dystrophy can affect the craniofacial growth pattern. Materials and methods The research was conducted on a sample of 27 patients with Steinert’s myotonic dystrophy (study group). Each subject underwent a clinical examination with impression-taking and intra- and extraoral photographs. A latero-lateral projection teleradiography in the mirror position was also taken and a cephalometric examination was performed. The assessed values were compared with those obtained from a group of healthy subjects (control group). Results Statistical analysis of the data obtained from the myotonic patients who developed the disease during the growth phase revealed alterations in the transversal plane and, to an even greater extent, the vertical one, with a high frequency of anterior open bite. Discussion and conclusions Regarding the pathogenesis of these types of skeletal dysplasias, the authors hypothesise a posterior rotation growth pattern, resulting from gravitational force prevailing over the deficit of the elevator muscles.

Key words: Myotonic dystrophy; Craniofacial growth; Orofacial musculature.

Introduction

The correlation between muscle function and craniofacial growth has been extensively investigated by clinical and experimental studies [Kiliaridis S., 1995] [Ueda S. et al., 1998] [Katsaros C., 2001] [Kiliaridis S. et al., 1989].

Facial musculature can affect growth of the maxillofacial region via a twofold mechanism: firstly, bone formation at the insertion of the muscle fascias results from activity of the muscle itself; secondly, muscle is one of the main components of the soft tissue matrix that, in normal conditions, guides the development processes of the splanchnocranium. The partial loss of muscular function, characteristic of several pathological patterns [Pearson A.M. et al., 1993], may account for alterations in normal craniofacial skeletal growth [Proffitt W.R., 2001]. Therefore, it is easy to see how the different myopathic processes that may arise during skeletal development play an important role in determining the subject’s growth pattern.

In patients with progressive muscular dystrophy, a malocclusion characterised by anterior open bite was first described by Brown and Losch, followed by White and Sackler. In both studies, however, the observations were limited to dentoalveolar alterations.

Studies on the correlations between muscular dystrophy and craniofacial development alterations have been conducted by a number of authors.

Kreiborg et al. [1978] described a case of congenital muscular dystrophy in which the cephalometric examination highlighted marked vertical facial development with extreme mandibular posterior rotation, back- and downward mandibular growth direction, and anterior dental open bite. In a study of patients with Steinert’s myotonic dystrophy and Duchenne muscular dystrophy, Kiliaridis et al. [1998] reported the prevalence of dysgnathic patterns, characterised by anterior open bite, posterior cross bite, increased overjet, narrow and deep palate roof, altered lingual posture and overeruption of the posterior teeth.

The development of malocclusion in patients with muscular dystrophy is strictly correlated with aberrant vertical craniofacial growth, which leads to alterations in head length, head breadth, interzygomatic distance, nose breadth, maxillary arch widths, palate depth, anterior and posterior face height, cranial base length, cranial base angles and other cephalometric measures [Staley et al., 1992].

Electromyographic studies on muscular dystrophy patients [Odman et al., 1996] show a considerable reduction in masticatory activity, which represents the predominant cause of the typical craniofacial alterations observed in these subjects. The main
muscles involved are the masseter and the anterior part of the digastric, since their muscular fascias have the highest positive correlation with vertical craniofacial growth [Ueda et al., 1998].

In a magnetic resonance study, Zanotelli et al. [2002] hypothesised that the altered masticatory function that causes changes in mandibular biomechanics is responsible for the TMJ alterations that are often seen in muscular dystrophy patients. The morphologic changes of the articular bone heads account for the most frequent TMJ alterations, although morphologic discal alterations are also frequently observed.

A study conducted by Engval et al. [1997] showed that patients with muscular dystrophy exhibit remarkable susceptibility to caries compared to healthy subjects; this is essentially due to a higher oral clearance of dietary sugar intake, reduced salivary secretion and lower oral hygiene levels, with subsequent higher plaque indexes [Balasubramaniam et al., 2008].

Steinert’s myotonic dystrophy is the most frequent form of muscular dystrophy, with an overall incidence of 1:20.000 in Caucasian populations [Bouhour et al., 2007]. It is a hereditary disease, caused by an alteration of a locus of the chromosome 19q13-2 and with an autosomal dominant transmission (meaning that the DNA alteration involves a single element of a pair of chromosomes), and is characterised by multisystemic clinical involvement [Friedman et al., 1980], [Dalal et al., 1972].

Muscular involvement is characterised by progressive hypotrophy and hypostenia of the facial muscles and of the semidistal and distal limb segments, associated with muscle decontraction difficulties (myotonic phenomenon) and muscle weakness, leading to an expressionless face and severe palpebral ptosis (Fig. 1), determining the characteristic feature called myopathic facies [Hellmuth et al. 1982].

Other typical signs are cataracts, heart-conduction and heart-rate anomalies (atrioventricular conduction defects, ventricular and supraventricular arrhythmias), endocrine system disorders (diabetes, dysthyroidism, fertility disorders, etc.), mild or moderate mental retardation [Modoni et al., 2008], intestinal motility disorders [Harper, 1989], and eye movement disorders [Osanai et al., 2007].

Age at the onset of symptoms and the severity of clinical manifestations tend to vary. In the typical form, the onset of muscle deficit appears in the second or third decade of life, usually with a slow progression; the early forms present a more severe symptomatic pattern, and are often associated with pulmonary complications and mental deficits [Turpin et al., 1980]. However, the life expectancy of Steinert’s disease patients is often lowered by lethal heart complications that, together with the bronchopulmonary infections, represent the most frequent cause of death [Souayah et al., 2007]. Myotonic dystrophy also leads to oropharyngeal dysphagia [Chiappetta et al., 2001] and increased oro-nasal resistance, probably due to alterations in the elastic properties of the pulmonary parenchyma [Fodil et al., 2004].

Materials and methods

The study was conducted on a sample of 27 patients (15 F, 12 M) ranging in age from 12 to 30 years and with Steinert’s myotonic dystrophy (study group), enrolled at the 2nd Neurological Clinic of the G. Martino University Hospital in Messina, Italy. Scientific Protocol has been authorized by the Ethic Commission of the Medical Faculty.

Each patient in the study group underwent clinical and instrumental examination at the Dental Clinic of the Orthodontic Department of the University of Messina. The clinical examination was complemented with cast analysis and intra- and extraoral photographs. Teleradiography in norma lateralis acquired in self-balance position was also taken, and the related cephalometric analysis performed.

The parameters considered for the study were as follows: ANB angle, SNB angle, Witts index, intermaxillary angle, craniomandibular angle, gonial angle, Sor-SNA distance, Sor-Me distance, and the difference between the two recorded values. The study group was divided into 2 subgroups according to the time of disease onset. Group 1-A, composed of 13 infant patients (7 M, 6 F), included subjects whose onset dated back to a dynamic developmental phase; Group 1-B, composed of 14 young adults patients (5 M, 9 F), included subjects whose disease manifested itself in a phase when growth could be considered completed [Hellmuth Muller et al., 1982].

The cephalometric values assessed for the study group were then compared with those obtained from a group of 30 healthy subjects (19 F, 11 M), ranging in

![Fig. 1 - Myopathic facies.](attachment:myopathic-facies.png)
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Table 1 - Descriptive statistics of the cephalometric parameters.

<table>
<thead>
<tr>
<th>Study Group 1-A</th>
<th>Myotonic patients with dystrophy onset before skeletal development completion</th>
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<tbody>
<tr>
<td>Mean</td>
<td>85</td>
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<tr>
<td>SD</td>
<td>3</td>
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<thead>
<tr>
<th>Study Group 1-B</th>
<th>Myotonic patients with dystrophy onset after skeletal development completion</th>
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<tbody>
<tr>
<td>Mean</td>
<td>83</td>
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<tr>
<td>SD</td>
<td>5</td>
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<tr>
<th>Control Group</th>
<th>Healthy subjects at growth completion with harmonious facial bones proportions</th>
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<tbody>
<tr>
<td>Mean</td>
<td>84</td>
</tr>
<tr>
<td>SD</td>
<td>3.2</td>
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</tbody>
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Results

Table 1 shows the descriptive statistics (mean and SD) of the cephalometric parameters considered for the study. The following data emphasise marked vertical facial growth in the study group subjects, characterised by mandibular posterior rotation and a tendency towards maxillary retrusion (Fig. 2).

The cephalometric examinations of Group 1-A patients showed a high prevalence of malocclusions, characterised by increased craniomandibular, intermaxillary and gonial angles, increased antero-inferior facial height and a tendency towards skeletal Class II. Significant statistical correlations were observed between the sagittal plane discrepancy and mandibular divergency (p<0.05), and between the antero-inferior facial height and mandibular divergency (p<0.01). Group 1-B patients did not show uniform or statistically significant skeletal alterations. The cephalometric examinations of these patients showed infact a tendency towards skeletal Class I, craniomandibular, intermaxillary and gonial angles, with values close to normal ones rather than Group 1-A patients.

Discussion

The clinical examinations of Group 1-A patients...
showed a prevalence of anterior open bite (8 subjects), posterior cross bite (11 subjects), mandibular plane steepness (13 subjects) and labial incompetence (9 subjects.). These patients exhibit a fairly typical appearance, clearly characterised by a long face. Group 1-B patients did not show uniform or statistically significant skeletal alterations.

Data analysis of the dystrophic patients whose disease onset dated to the developmental phase of the maxillofacial growth shows a nearly constant and statistically significant increase in the cephalometric parameters that define skeletal dysplasias with vertical excess on the horizontal plane.

For these patients, one can hypothesise that hypotrophy and muscle deficit lead to greater susceptibility to the effects of gravitational force, with subsequent clockwise mandibular rotation. Mandibular posterior rotation would lead to stretching of perifacial soft tissues, molar overeruption and positional modifications of the tongue.

This pathophysiological pattern explains the set of alterations found in this study.

The reduction in palate transverse diameters may derive from the centripetal forces that develop due to stretching of the perifacial soft tissues and the anomalous position of the tongue, which is unable to exert its physiological morphogenetic effect on the palatine roof.

The ogival palate frequently observed in these subjects can be explained by the excessive dentoalveolar overeruption of the posterior sectors.

The inclination of the upper incisors is not altered, which is mainly due to the minimal muscle activity of the upper lip. The lower incisors, on the contrary, are always lingually inclined. This feature is presumably correlated with the reduced muscle tone of the tongue.

Obviously the aberrant growth pattern, specially for the patients with dystrophy onset before skeletal development completion, affects functions such as chewing, swallowing and speech.

Early diagnosis would be helpful for this patients, because early treatment within functional appliances, could reduce the severity of the malocclusion described above.

Conclusions

For patients with disease onset before the facial bones have fully developed, the statistically significant alterations of the anthropometric parameters that are observed demonstrate the crucial role of proper muscle function in the harmonious development of facial bones. Therefore, we can affirm that Steinert’s muscular dystrophy, as well as the different myopathic processes that may arise during the skeletal development, play a crucial role in determining the patient’s craniofacial developmental pattern.

References


