Muscle function and craniofacial morphology: a clinical study in patients with myotonic dystrophy

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SUMMARY The occlusal traits and the craniofacial morphology were studied in patients with an altered muscle function caused by myotonic dystrophy. Twenty-four adult patients were examined and compared with a matched group of healthy individuals.

The condition of the masticatory muscles was evaluated by measuring the maximal bite force. The dental arches and the occlusal traits were analysed on dental casts. Lateral cephalograms were taken in the patients with myotonic dystrophy to study the craniofacial morphology.

It was found that the patients suffering from myotonic dystrophy had weak masticatory muscles, which might be caused by the disease. A high prevalence of malocclusions (postnormal occlusion, anterior open bite and lateral cross bite) was found among these patients. Their craniofacial morphology showed a vertical aberration, characterized by a large angle between the mandibular and palatal planes and a steep mandible. These findings seem to be most pronounced in patients with an early onset of the disease and support the hypothesis that reduced muscle function may cause changes in the craniofacial morphology.

Introduction

Clinical studies have shown a correlation between the bite force and facial morphology (Ringqvist 1973, Ingervall and Helkimo 1978, Proffit et al. 1983). Subjects with a strong bite force had a more anteriorly inclined mandible, smaller anterior and greater posterior facial heights, as well as a smaller gonial angle, in contrast to those with a weak bite force, who had longer anterior and shorter posterior facial heights and a larger gonial angle. These findings were used to support the theory that the form of the face partly depends on the strength of the muscles. However, according to Throckmorton et al. (1980), the low bite force in persons with long faces might be a result of the geometric arrangement of the lever system of the jaw. This means that the elevator muscles of the jaw are in an unfavourable mechanical position when the gonial angle is obtuse and the mandibular plane angle is steep. Thus, according to this view, it is the muscular function which is influenced by the skeletal form.

Animal experimental models have been used with the purpose of elucidating the relationship between craniofacial morphology and muscle function. These models were based on changes in the function induced by removing the masticatory muscles (Horowitz and Shapiro 1951, 1955, Moss and Meehan 1970), disrupting the occlusion (MacNamara 1973, Petrovic et al., 1982, Tonge et al. 1982, Woodside et al. 1983), or changing the consistency of the food (Watt and Williams 1951, Beecher and Corruccini 1981, Kiliaridis 1986).

The skeletal changes induced by alteration of muscle function indicate that masticatory muscle function has a significant influence on craniofacial growth. Although the results of these animal experimental studies elucidate the basic mechanism of bone remodelling and muscle function, they can not be applied to man without reservations.

Myotonic dystrophy is a muscular disease of autosomal dominant inheritance. The progress of the disease is, in general, extremely slow and the muscles involved become atrophic and weak. Myotonic dystrophy has a primary distal distribution, the principal affliction being of the hands, forearms, lower legs and the muscles of the jaws, neck, face and eyelids. The course of the disease varies from individual to individual. The disease may involve the muscles very early...
in life, but the development of severe muscle weakness can appear much later, entailing a late diagnosis (Harper 1979).

The facial appearance in patients with myotonic dystrophy is characteristic (Fig. 1), related to the weakness of the muscles in the face and the jaws. However, the possible effect of the reduced functional capacity of the masticatory muscles on the craniofacial skeleton has not yet been studied.

Thus, the aim of the present study was to investigate the occlusal traits and craniofacial morphology in patients with an altered muscle function caused by myotonic dystrophy.

Subjects and methods
Twenty-seven adult patients were examined, 18 woman and 9 men, all registered at the East Hospital in Göteborg with a diagnosis of myotonic dystrophy. Three of the patients wore dentures, and were therefore excluded from this study. The remaining 24 patients had each at least 9 pairs of opposing teeth. The average age of the 16 female patients was 42.3 years (s.d. 10.1, range 30-62) and of the 8 male patients 40.5 years (s.d. 9.0, range 28-61).

The degree and the onset of the disease varied from patient to patient. From the medical records it was found that the group of male patients were more seriously affected by the disease than the female group. The patients were compared with a group of healthy individuals who were visiting a Public Dental Health Clinic for their annual dental check-up. This control group was matched to the group of myotonic dystrophy patients in respect of age, sex and number of occluding pairs of teeth.

Impressions were taken in order to construct dental casts for dental analysis of the occlusal traits. The relation between the upper and lower jaws was recorded in the intercuspal position. The dimensions of the palatal vault were measured on the casts (Fig. 2) The palatal height was measured from the highest point of the palatal...
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Figure 2  The dimensions of the palatal vault measured on the casts. H: palatal height, W: palatal width. The shape of the palatal vault was estimated by the ratio H/W*100.

Figure 3  Cephalometric reference points and part of the lines used in the present study (Definitions in Hasund, 1985).

vault perpendicular to the occlusal plane at the second premolars, and the palatal width was measured between the palatal surfaces of the second premolars. In order to avoid individual size variation, the shape of the palatal vault was estimated by the ratio H/W*100.

The number of occluding pairs of teeth was counted and divided by the existing pairs of teeth i.e. the pairs of teeth with or without contact between the antagonists, in order to calculate the ratio of the functioning pairs of teeth in the dental arches.

The condition of the masticatory muscle was evaluated by measuring the maximal bite force in the region of the second premolars and first molars, using a biting fork equipped with a strain gauge (Helkimo et al. 1975).

Lateral cephalograms were taken of patients with myotonic dystrophy to study the craniofacial morphology. The reference points and lines used in cephalometric analysis are shown in Fig 3. The controls were not exposed to x-rays for ethical reasons. Therefore, the results obtained from the cephalometric analysis of the patients will be compared with the normal standards for the facial skeleton of Swedish healthy adults, based on 19 female and 11 male 30 year-old individuals (Thilander et al., 1982, Persson and Thilander 1987).

Error of the methods
The error of measurements in the dental analysis of the stone cast and the cephalometric analysis was calculated after duplicate determinations, based on recordings of 15 randomly chosen subjects. The bite force was recorded twice in a group of 15 healthy individuals within a two-week period.

The error of the method (Se) was calculated by the formula

\[ Se = \sqrt{\frac{\Sigma d^2}{2N}} \]

where d is the difference between the first and the second recordings and n is the number of double determinations or pair recordings. The error of measurements did not exceed 0.5 mm in the dental cast analysis and 1 degree in the angular measurements and 1 mm in the linear measurements of the cephalometric analysis. The error of the method for the maximal bite force recordings was 54 N.
Statistical methods

The non-paired t-test was used to assess significant differences between the groups regarding continuous variables. For non-continuous variables the $\chi^2$ test was used to test the significance of differences in frequencies between the groups.

Results

The analysis of the dental casts revealed a high frequency of malocclusions among the patients with myotonic dystrophy (Table 1).

Almost half of the myotonic dystrophy patients (46%) had a postnormal occlusion, which was higher (p<0.07) than found in the healthy individuals (21%). The frequency of patients with an increased horizontal overjet (38%) and frontal open bite (33%) in the myotonic dystrophy group was significantly higher than in the healthy controls, where these malocclusions had a frequency of 8 percent each.

Unilateral or bilateral cross bite was found in half of the patient group, while in the matched group the frequency was 29 per cent.

The number of occluding pairs of teeth in relation to those existing in the arches, with the antagonists not necessarily in contact, was found to be 71 percent (SEM:4) in the group with muscular dystrophy, being significantly lower (p<0.001) than in the control group (92%, SEM:2).

The measurements of the dimensions of the palatal vault showed that the patients suffering from myotonic dystrophy had a deeper and narrower palatal vault, both in absolute and relative values (Table 2).

The cephalometric analysis (Table 3) showed a retrognathic facial type among the females, while among the males there was no statistically significant difference in the anteroposterior skeletal relationship between the myotonic dystrophy patients and healthy normal individuals. However, the craniofacial morphology of the patients studied was characterized by differences in the vertical direction (Fig. 4) with a high mandibular-palatal planes angle (ML/NL) and a large gonial angle (Gn-tgo-Ar), in both men and women. The patients had a long lower anterior facial height (Sp'-Gn) (Table 3).

The maximal bite force was found to be significantly lower in the myotonic dystrophy patients, being on average less than the half of the mean value of the matched healthy controls (Table 4).

Discussion

These observations were made in a group of patients whose disease varied in duration and...
Table 3 Cephalometric variables of myotonic dystrophy patients and normal standards (Persson and Thilander 1987). (ns = not significant, * = p≤0.05, ** = p≤0.01, *** = p≤0.001).

<table>
<thead>
<tr>
<th></th>
<th>Females</th>
<th></th>
<th>Males</th>
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</thead>
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<tr>
<td></td>
<td>Myotonic dystrophy</td>
<td>Normals</td>
<td>Myotonic dystrophy</td>
<td>Normals</td>
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<tr>
<td></td>
<td>(n = 16)</td>
<td>(n = 19)</td>
<td>(n = 8)</td>
<td>(n = 11)</td>
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<tr>
<td>SNA</td>
<td>81.2 ± 3.8</td>
<td>84.2 ± 3.0</td>
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<td>81.5 ± 4.5</td>
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<td>SNB</td>
<td>77.3 ± 5.2</td>
<td>81.4 ± 2.3</td>
<td>**</td>
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<td>ANB</td>
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<td>2.8 ± 2.0</td>
<td>ns</td>
<td>1.9 ± 1.6</td>
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<tr>
<td>SNPg</td>
<td>78.2 ± 5.4</td>
<td>83.0 ± 2.2</td>
<td>**</td>
<td>80.6 ± 3.6</td>
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<td>ML-NSL</td>
<td>34.8 ± 8.8</td>
<td>26.4 ± 4.8</td>
<td>**</td>
<td>37.3 ± 7.1</td>
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<td>NL-NSL</td>
<td>6.4 ± 2.9</td>
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<td>ns</td>
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<td>ML-NL</td>
<td>28.4 ± 8.8</td>
<td>21.2 ± 4.8</td>
<td>**</td>
<td>31.5 ± 7.3</td>
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<td>NSBa</td>
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<td>Gn-nto-Ar</td>
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<td>117.3 ± 5.6</td>
<td>**</td>
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<tr>
<td>(isa-is)-(ia-ii)</td>
<td>131.7 ± 13.0</td>
<td>134.6 ± 6.0</td>
<td>ns</td>
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<td>(isa-is)-NA</td>
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<td>18.5 ± 4.3</td>
<td>ns</td>
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<td>is-NA mm</td>
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<td>3.4 ± 1.6</td>
<td>ns</td>
<td>5.8 ± 2.0</td>
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<td>ii-NB mm</td>
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<td>ns</td>
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<td>Pg-NB mm</td>
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<td>N-Sp' mm</td>
<td>52.3 ± 2.8</td>
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<td>Sp'-Gn mm</td>
<td>72.0 ± 7.2</td>
<td>67.6 ± 4.5</td>
<td>*</td>
<td>80.3 ± 4.9</td>
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<td>N-Sp/Sp'-Gn</td>
<td>0.72 ± 0.08</td>
<td>0.78 ± 0.07</td>
<td>*</td>
<td>0.70 ± 0.05</td>
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</table>

Figure 4 Mean facial diagrams illustrating the differences in facial morphology between the patients with myotonic dystrophy and health normal individuals. (a) Females and (b) Males.
Table 4  Maximal bite force (N) in 24 myotonic dystrophy patients and 24 matched controls; 16 females and 8 males in each group. (** = p<0.001).

<table>
<thead>
<tr>
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<th>Myotonic dystrophy</th>
<th>Controls</th>
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</thead>
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<td>Maximal bite force</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Females</td>
<td>138.2 79.4</td>
<td>383.2 130.3</td>
</tr>
<tr>
<td>Males</td>
<td>204.8 58.8</td>
<td>540.0 137.2</td>
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degree. In male patients, who were more seriously affected by the disease than females, the vertical aberration of the craniofacial morphology and the occlusal anomalies seem to be more pronounced.

In the patient group, the frequency of malocclusions was very high when compared with the matched group of healthy individuals or with previous epidemiological studies in adults (Ingervall et al, 1978, Mohlin 1982). Furthermore, the masticatory muscles in these patients had been affected by the disease, as indicated by the low biting force compared with that in the healthy controls or with that found in previous studies (Helkimo et al, 1975).

The involvement of these muscles may have caused a lowering of the mandible, either due to gravitation or due to the activity of possibly less involved suprathyroid muscles (Kreiborg et al, 1978). Lowering of the mandible can, in turn, affect the tongue position and head posture. Thus, a new situation is established around the teeth transversely. The lowered tongue is not in a position to counterbalance the forces developed during the lowering of the mandible by the stretched facial musculature. This may affect the position of the teeth transversely, decreasing the width of the palate and causing a crossbite.

The lowered position of the mandible, in combination with the decreased biting forces, may permit an over eruption of the teeth (Proffit 1978). In this case, the palatal vault height is possibly increased because of the over eruption, and the mandible rotates posteriorly, causing an increased angle between the mandibular and the palatal planes. The increased frequency of Angle Class II malocclusion among the myotonic dystrophy patients may be attributable to the posterior rotation of the mandible (fig. 5).

The increased gonial angle of the mandible means that the form of the mandible was changed, since the tension applied on the bony structures by the masticatory muscles was decreased. This is in accordance with our previous experiments in which the angle of the mandible was decreased in animals with a low masticatory function, owing to a soft diet (Kiliaridis et al., 1985, Kiliaridis and Shyu 1988). The changes in the gonial angle of the mandible are in accordance with Wolff's law (1892) that the shape and the structure of the bones depend on the stress placed upon them by the functional forces of the muscles.

The facial and occlusal characteristics of the patients suffering from myotonic dystrophy are reminiscent of the 'adenoid face' (Linder-Aronson 1970). In the adenoid face the neuromuscular adaptation causes a secondary lowering of the mandible and a change in head posture because of the need to facilitate mouth-breathing (Harvold 1983). However, in myotonic dystrophy it is the weakness of the elevator muscles which causes the lowering of the mandible and permits mouth-breathing, as the patients had no obstruction of nose-breathing. The fact that the skeletal changes in both cases are similar can be easily explained by the alteration in the muscular function, rather than the changes in air passage. This is in agreement with Harvold (1983) who stated that 'changes in muscle activity, such as muscle recruitment for oral respiration, may cause remodelling of the jaws and produce a malocclusion. It should be noted that the crucial factor is a consistent change in muscle use and not a change in airflow...'.

In conclusion, patients with myotonic dystrophy showed weak masticatory muscles. Their craniofacial morphology was characterized by a large angle between the mandibular and palatal planes and a steep mandible. Malocclusions found frequently among these patients were postnormal occlusion, anterior open bite and lateral cross bite. These findings seem to be most pronounced in patients with an early onset of the disease and support the hypothesis that reduced muscle function may cause changes in the craniofacial morphology. Myotonic dystrophy may be one of the aetiological factors in 'long face syndrome'. Since the disease has an extremely slow progress, in certain cases the vertical aberration in the craniofacial growth may be one of the initial signs of the disease.
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