Craniofacial morphological differences between Down syndrome and maxillary deficiency children

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SUMMARY Maxillary deficiency is one of the facial features of Down syndrome (DS). Differences in craniofacial morphology between DS and nonsyndromic skeletal Class III malocclusion with maxillary deficiency remain unclear. This study compared the craniofacial differences of white male children from Central–Western Brazil with DS (n = 30, mean age: 8 years 3 months), skeletal Class III profile with maxillary deficiency (n = 30, mean age: 7 years 9 months), and skeletal Class I profile (n = 30, mean age: 8 years 2 months), using lateral cephalometric radiographs. The differences among the three groups were compared with analysis of variance and Tukey’s tests. The DS group showed reduced anterior cranial base (S–N, P < 0.001) and facial dimensions (Co–Gn, N–Me, N–ANS, and ANS–Me, P < 0.001), except in posterior dimensions (S–Go, P < 0.005; Ar–Go, P > 0.005). Maxillary length (Co–A, P < 0.001) and facial convexity (NAP, P < 0.005) were reduced when compared with the control group, although maxillary position to cranial base (SNA, P < 0.005) was within the normal range. A flattened cranial base (BaSN, P < 0.001) also contributed to differentiating DS from nonsyndromic groups. The group with maxillary deficiency showed a more unfavourable maxillomandibular relationship (MMD, P < 0.001) and a mandibular protrusion (SNB, P < 0.001). Subjects with DS differed from Class III with maxillary deficiency with respect to the flatter cranial base and reduced maxillary length. Maxillary deficiency was not so expressive in the face of DS subjects because of the overall reduction in craniofacial dimensions.

Introduction

Down syndrome (DS) is the most prevalent genetic malformation (1 in 750–800 live births) and was first described in 1866 by the English physician Langdon Down (Frostad et al., 1971; Fink et al., 1975; Silva et al., 1997). In 1959, Lejeune et al. identified a chromosomal numerical change, with karyotypes characterized by trisomy 21 (Lejeune et al., 1959). DS has three genetic etiologies that are expressed in phenotypes with greater or lesser syndrome characteristics. The 21 regular trisomy is the most common (94.1 per cent), while translocations (3.6 per cent) and mosaics (2.3 per cent) occur in a smaller proportion (Stoll et al., 1998).

The complex genetic disorder is clinically characterized by a combination of mental and physical features (Frostad et al., 1971; Fink et al., 1975; Silva et al., 1997). Although a wide spectrum of phenotypes can be expressed, typical craniofacial features make DS distinct and easily recognizable. Several studies have described the craniofacial morphological characteristics of DS, from prenatal to postnatal periods (Gosman, 1951; Austin et al., 1969; Cohen et al., 1970; Jensen et al., 1973; Fink et al., 1975; Westerman et al., 1975; Jara et al., 1993; Farkas et al., 2001; Guichard-Costa et al., 2006). Questionnaire-based examination (Oliveira et al., 2008), clinical examination (Shapira et al., 2000; Oliveira et al., 2008), computerized tomography (Ferrario et al., 2005), anthropometrics (Farkas et al., 2001), dental cast (Westerman et al., 1975; Shapira et al., 2000), photographs (Gosman, 1951), radiographs (Austin et al., 1969; Shapira et al., 2000), magnetic resonance imaging (Guimaraes et al., 2008), and cephalometric (Frostad et al., 1971; Fink et al., 1975; Motohashi, 1985; Fischer-Brandies et al., 1986; Fischer-Brandies 1988; Quintanilha et al., 2002; Suri et al., 2010) studies of DS have all pointed out reduced head and facial dimensions; brachycephalic cranium; shorter and flatter cranial base; reduced or absent frontal sinus and nasal bone; hypotelorism; epicantthic fold; seal-like nose; small ear; insufficient lip seal; high palate with reduction in its length; narrowed oropharynx; presence of mouth breathing and obstructive sleep apnoea; normal or reduced goniac angle; hypotonic and protrusive tongue; relative rather than absolute macroGLOSSIA; delayed dental eruption; alterations in the number (hypodontia), dimensions, shape, and position of the teeth; and reduced, normal, or prognathic mandible and midface deficiency with reduced nasal protrusion. Craniofacial dysmorphology and muscular imbalance predispose to a higher prevalence of malocclusion, with special emphasis on anterior (Jensen et al., 1973; Fischer-Brandies, 1988; Borea et al., 1990; Oliveira et al., 2008) and posterior (Janson et al., 2009) open bite, posterior crossbite (Gosman, 1951; Jensen et al., 1973; Oliveira...
et al., 2008), dental protrusion (Gosman, 1951), crowding (Ondarza et al., 1993), and pseudo and skeletal Class III malocclusion (Gosman, 1951; Cohen et al., 1970; Jensen et al., 1973; Fischer-Brandies, 1988; Borea et al., 1990). Skeletal Class III malocclusion exhibits lesser facial convexity as a result of maxillary deficiency, mandibular protrusion, or both. In general, maxillary deficiency, also called ‘retrognathic maxilla’, is a common finding in these subjects (Cohen et al., 1970; Fink et al., 1975; Ferrario et al., 2005).

Technological advances in diagnoses, treatment protocols, special education programs, as well as an increasing effort to integrate these individuals into society have helped in giving these disabling individuals a better quality of life as well as a longer one (Fischer-Brandies et al., 1986; Borea et al., 1990; Ingerval and Schmoker, 1990; Becking and Tuinzing, 1991; Glatz-Noll and Berg, 1991; Wolford and Cottrell, 1996; Carlsted et al., 1999, 2003; Desai and Flanagan, 1999). It is accepted that the way in which the maxilla and the mandible grow and develop is crucial to facial composition, and this has considerable impact on the lives of these people because it causes problems in their daily activities, including self-esteem and social acceptability issues.

Craniofacial characteristics of DS children have been previously studied (Fischer-Brandies, 1988). A number of reports have suggested that maxillary complex deficiency is recognized as one of the facial features (Fischer-Brandies et al., 1986; Fischer-Brandies, 1988). However, as head and facial dimensions are overall reduced when compared with normative values, doubts linger about the differences between DS and decreased maxillary jaw size subjects. This direct comparison has never been described and it remains unclear. Moreover, concerns about facial appearance in DS are continuously increasing and this information may provide information to assist in treatment planning.

The aim of the present study is to clarify this difference by comparing the craniofacial complex characteristics between DS and nonsyndromic skeletal Class III malocclusion with maxillary complex deficiency in a sample of homogeneous male children from Central–Western Brazil.

Subjects and methods

The study was approved by the Ethical Committee of the Medical Faculty of Federal University of Goiás (022/2001), according to the Brazilian Health Ministry no. 196/96 resolution. Informed consent was obtained from at least one parent or legal guardian.

Sample selection

This study included three groups of white male children from Central–Western Brazil with mixed dentition: a DS group, a skeletal Class III malocclusion with maxillary deficiency group, and a control group with skeletal Class I malocclusion. All of them were descendents of Brazilian parents and grandparents, and none had undergone craniofacial trauma or surgery, orthodontic procedures, or hormonal growth therapy. The male children were used because they were more often found mainly among the institutionalized DS individuals.

The syndromic group comprised of 30 DS children. The mean age was 8 years and 3 months (range: 6 years and 6 months to 11 years and 9 months). The diagnosis of DS was confirmed through pre-existing karyotype examination. The sample was prospectively selected from the patient roster at the Faculty of Dentistry of the Federal University of Goiás as well as from the teaching institutions for the disabled subjects (APAE, CRESPA, and Pestalozzi Institute) in Goiânia, state of Goiás, Brazil. Cephalograms were taken on the DS group in the same radiologic appliance.

The maxillary deficiency group comprised of 30 pretreatment subjects with a mean age of 7 years and 9 months (range: 5 years and 9 months to 10 years and 4 months) for whom cephalometric radiographs were taken. The inclusion criteria for this study group were nonsyndromic skeletal Class III malocclusion with maxillary deficiency and a small shift between centric occlusion and occlusion in centric relation. The selection was based on frontal and lateral photographs, according to the criteria cited by Staudt and Kiliaridis (2009), with the addition of marked deficiency in the paranasal region.

The control group comprised of 30 subjects with a mean age of 8 years and 2 months (range: 6 years and 6 months to 11 years and 5 months) and who had their cephalometric radiographs taken. This group presented a Class I malocclusion in mixed dentition and well-balanced faces on frontal and lateral photographs. Both the groups of nonsyndromic samples were retrospectively selected at random from a private orthodontic practice and the Faculty of Dentistry of Federal University of Goiás, Goiânia, State of Goiás, Brazil.

Cephalometric assessment

The cephalometric radiographs for all subjects were taken using the same cephalostat and machine with an enlargement factor of the cephalostat of 1.0645 (6.45 per cent; Panoura 10 CSU, The Yoshida Dental MGF Co., Tokyo, Japan). The children were in standing position and adequately protected, with the teeth in centric occlusion, lips in relaxed position, and Frankfurt plane parallel to the floor. Radiographic films were developed in an automatic processor (Flat Co., Japan). The radiographic procedure was specially complicated with the DS group. The 30 selected radiographs of DS children were of sufficient quality and around 6 radiographs were excluded due to bad quality of images. There were initially more subjects in the DS group, but for ethical reasons, the imperfect radiographs were excluded from the initial sample and were not repeated for these subjects (Figure 1).
Craniofacial morphology was first evaluated with a traditional cephalometric procedure. Fifteen skeletal cephalometric landmarks (Figure 2A) were marked by hand on an acetate paper in a darkened room by the same investigator. The radiographic images were then digitized using a computer and software (Radiocef 2.0, Radio Memory, Brazil) to obtain 15 linear and 7 angular standard measurements (Figure 2B) based on those developed by Martins et al. (1998).

**Error analysis**

To determine the reliability of the cephalometric method, 15 randomly selected cephalometric radiographs from each group were traced and measured twice with a 2-week interval by the same investigator. Random error (S) was calculated according to Dahlberg’s formula: 

\[ S = \sqrt{\frac{\sum d^2}{2n}} \]

where \( d \) is the difference between the two measurements and \( n \) is the number of tested radiographs. Systematic errors were evaluated with paired t-tests at \( P < 0.05 \) (Houston, 1983).

**Statistical analysis**

The mean values and standard deviations were computed for all variables. The three groups were compared with a one-way analysis of variance followed by a post hoc test (Tukey’s honestly significant difference). The significance level was set at \( P < 0.05 \) (significant) and \( P < 0.01 \) (highly significant). SPSS 14.0 for Windows (SPSS Inc., Chicago III) was used for the statistical analysis.

**Results**

The errors’ study showed that only variables ANS perp OrPo and PNS perp OrPo had systematic errors (\( P < 0.05 \)), and the casual errors’ mean was 0.6 mm (range: 0.4–0.9 mm) for linear and 0.9 degrees (range: 0.5–1.1 degrees) for angular measurements. The cephalometric variables related to Frankfurt horizontal were not so reproducible because the two landmarks were positioned laterally to the mid-sagittal plane and thereby were prone to be distorted even by a slight tilt of the head in the cephalostat.

The angular and linear cephalometric measurements for the DS, maxillary deficiency, and control groups and the statistical difference among them are shown in Table 1. Fifteen of the 22 measurements showed a statistically significant difference between the studied groups \( (P < 0.05) \), with 13 demonstrating a more pronounced difference \( (P < 0.01) \). Craniofacial linear dimensions of DS were shorter than those observed in the other groups, especially the skull anterior base (S–N; \( P < 0.01 \)), maxillary length (Co–A; \( P < 0.01 \)), mandibular body length (Go–Gn; \( P < 0.01 \)), and anterior facial heights (N–Me, N–ANS, and ANS–Me; \( P < 0.01 \)). The maxillary deficiency group showed an intermediate linear dimension among the three groups, except for the mandibular body length (Go–Gn, \( P = 0.000 \)) and the maxillomandibular relationship (MMD, \( P < 0.01 \)), which were higher and exhibited more skeletal imbalance.

The skull base was flatter for the DS group, when compared with the others (BaSN, \( P < 0.01 \)). The reduced maxillary length in the DS and maxillary deficiency groups did not affect the degree of maxillary protrusion in relation to the cranial base (SNA and N-perp–A, \( P > 0.05 \)). Considering the Frankfurt horizontal plane (OrPo) as a reference, the vertical position in the anterior region and the degree of maxilla tilting did not show a statistically significant difference (ANS perp OrPo, OrPo.ANS–PNS, \( P > 0.05 \)).

The length of the mandible body was markedly smaller for DS (Go–Gn, \( P < 0.01 \)), whereas it was similar between the maxillary deficiency and the control groups. However, the position of the mandible in relation to the cranial base showed a statistically significant protrusion for the maxillary

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Figure 1  Cephalometric radiograph representative of Down syndrome (DS; A), skeletal Class III with maxillary deficiency (B), and control (C) groups at similar ages. Note that the facial dimensions of DS are reduced, when compared with the others.
CRANIOFACIAL MORPHOLOGICAL DIFFERENCES

Figure 2  (A) The cephalometric points. Landmark—S = sella, the centre of sella turcica; N = nasion, the most anterior point on the frontonasal suture; Or = orbitale, the lowest point on the infraorbital margin; Po = porion, the uppermost point of the body of the external auditory meatus, usually regarded as coincident with the uppermost point of the ear rods of the cephalostat; ANS = anterior nasal spine, the most anterior point of hard palate; intersection of anterior and upper maxilla part with nasal fossae floor. Point A = subspinale, the deepest midline point between the anterior nasal spine and the prosthion; Point B = supramentale, the deepest point in the bony outline between the infradentale and the pogonion; Pog = pogion, the anterior-most point of the bony chin; Me = menton, the lowest point on the bony outline of the mandibular symphysis; Gn = gnathion, the most anterior and inferior point on the body outline of the chin, situated at equidistance from Pog and Me; Go = goion, the most lateral external point at the junction of the horizontal and the ascending ramus of the mandible; Ar = articularis, the point of intersection of the dorsal contours of the posterior border of the mandible and the temporal bone; Co = condylium, the most upper and posterior point of mandibular condylion outline; Ba = basion, the most posterior and inferior point of magnum foramen anterior portion; PNS = posterior nasal spine, the most posterior point of hard palate; Plates—FH plane = Frankfort horizontal plane, a transverse plane through the skull represented by a line joining porion and orbitale. (B) Linear (mm) and angular (°) measurements according to localization—cranial base: BaSN (°) = cranial base angle; Ba–N (mm) = total cranial base length; Ba–S (mm) = posterior cranial base length; S–N (mm) = anterior cranial base length. Maxilla: SNA (°) = the antero-posterior position of the maxilla relative to the anterior cranial base; Co–A (mm) = maxillary effective length, distance between condylium and A point; N–perp–A (mm) = nasion perpendicular A point, the distance of A point to a vertical line existing downward from nasion, perpendicular to the FH plane; ANS perp OrPo (mm) = the smaller distance of ANS point to the FH plane; PNS perp OrPo (mm) = the smaller distance of PNS point to the FH plane; OrPo/ANS–PNS (°) = angle between the FH (OrPo) and the palatal plane (ANS–PNS). Mandible: SNB (°) = the antero-posterior position of the mandible relative to the anterior cranial base; OrPo/Go–Gn (°) = angle between the FH (OrPo) and the mandibular plane (Go–Gn); ArGoMe (°) = gonial angle—angle between the intersection of line across Ar and Go with Go and Me line; Co–Gn (mm) = mandibular effective length—distance between Co and G point. Facial convexity and maxillomandibular relation: NAP (°) = facial convexity angle—supplementary angle between N, A, and Pog points; ANB (°) = the magnitude of the skeletal jaw discrepancy angle between N–A and N–B lines, showing SNA and SNB angle difference; MMD = maxillomandibular differential—the ratio of effective maxillary length (Co–A) effective mandibular length (Co–Gn). Facial heights: N–Me (mm) = total anterior face height; N–ANS (mm) = anterior upper face height; ANS–Me (mm) = anterior lower face height; S–Go (mm) = total posterior face height; Ar–Go (mm) = posterior lower face height.

deficiency group (SNB, \( P = 0.002 \)) and similarly for the others probably affected by the length of the cranial base. Gonial angle was statistically similar among the groups (Ar,Go,Me, \( P = 0.050 \)).

Statistical analysis revealed that the facial convexity and MMD relationship was more significant for the maxillary deficiency group, whereas the DS group maintained an intermediate level of skeletal imbalance (NAP, \( P = 0.012 \); ANB, \( P = 0.002 \); MMD, \( P < 0.01 \)).

Anterior facial heights were significantly shorter in the DS group, when compared with the maxillary deficiency and control groups (N–Me, N–ANS, ANS–Me; \( P < 0.01 \)), but mandible ramus height, which refers to the posterior lower facial height, was similar for the three groups.

The data from this study demonstrate that significant craniofacial differences exist between DS children and nonsyndromic children with maxillary deficiency and balanced face (control). To visualize these differences, a schematic cephalogram was constructed and superposed based on the mean cephalometric measurement data (Figure 3).

Discussion

Consistent with the more common concept found in literature (Frostad et al., 1971; Fink et al., 1975; Motohashi, 1985; Fischer-Brandies et al., 1986; Fischer-Brandies, 1988; Quintanilha et al., 2002; Suri et al., 2010), the present study showed that linear dimensions of the skull base and face of DS were reduced, when compared with Class I skeletal profile (Figure 1). The control group was within the normal range established by another Brazilian study with balanced skeletal face for the same mean age (Martins et al., 1998). In addition, the present study also compared these groups with a maxillary deficiency sample, which assumed an intermediate linear craniofacial dimension among DS and control groups. However, the exception was for the longer mandibular length (Go–Gn), which
contributed to the poorer MMD relationship and made the facial profile appear more concave. Although maxillary length was more significant in DS, the overall reduced facial dimensions camouflaged the facial appearance with greater maxillary deficiency (Figure 3).

The DS sample presented slightly larger linear measurements, when compared with those reported by Fischer-Brandies et al. (1986) and Fischer-Brandies (1988), for the same mean age. Probably, these differences may be explained by the following reasons: racial features, different radiological techniques, slightly different localization of some landmarks, and homogeneity of the samples. The most numerically expressive DS samples were published by Fischer-Brandies et al. (1986; n = 970) and Fischer-Brandies (1988; n = 1896); although these studies are important, there is no reference to ethnicity and origin. The present study, though numerically limited (n = 30), was compensated with a homogeneous group of subjects of same gender, age, and ethnicity.

According to Fischer-Brandies et al. (1986), the craniofacial anomaly is already present at birth and becomes more severe with increasing age. In the present study, because of the limited sample, it was not possible to divide the sample by age. However, efforts were taken to establish a homogeneous sample that represented white children from Central–Western Brazil. Therefore, only one gender was selected. The low prevalence of DS in the general population led the authors to contact institutions for the disabled persons. The availability of a higher number of male children in these places led to the decision to study only males.

Many problems developed when trying to obtain cephalometric radiographs in the DS group. The children were uncomfortable with the metallic porion, it was difficult to keep them still long enough to take the exposure, and extensive preconditioning was necessary to obtain the perfect head position. However, some children, even with the combined efforts of parents and the investigator, refused to be radiographed.

The presence of the syndrome itself is an important factor in craniofacial variation. The shorter anterior cranial base in DS may be related to the smaller size of the brain (Guirand-Costa et al., 2006). The angle of cranial base deflection was typically greater in the syndromic group (BaSN = 134.6

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### Table 1  Cephalometric measurements in the Down syndrome (DS; n = 30), skeletal Class III with maxillary deficiency (n = 30), and control (n = 30) groups

<table>
<thead>
<tr>
<th>Variable</th>
<th>DS Mean (SD)</th>
<th>Maxillary deficiency Mean (SD)</th>
<th>Control Mean (SD)</th>
<th>p&lt;sup&gt;1&lt;/sup&gt;</th>
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<tbody>
<tr>
<td>Cranial base</td>
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<tr>
<td>BaSN (°)</td>
<td>134.6 (4.90)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>126.4 (5.60)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>128.1 (2.85)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>Ba–N (mm)</td>
<td>96.6 (5.28)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>97.5 (3.99)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>103.3 (5.86)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
</tr>
<tr>
<td>Ba–S (mm)</td>
<td>42.2 (3.08)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>42.9 (2.62)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>44.2 (2.84)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.050</td>
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<tr>
<td>S–N (mm)</td>
<td>62.2 (3.78)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>65.8 (3.35)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>69.6 (4.22)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>Maxilla</td>
<td></td>
<td></td>
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<tr>
<td>SNA (°)</td>
<td>79.9 (3.91)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>81.2 (4.56)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>80.9 (3.19)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.448</td>
</tr>
<tr>
<td>Co–A (mm)</td>
<td>75.4 (4.56)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>79.7 (3.59)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>84.8 (4.36)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>N–perp–A (mm)</td>
<td>−1.0 (2.36)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>−1.6 (3.67)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>−0.4 (3.45)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.415</td>
</tr>
<tr>
<td>ANS perp OrPo (mm)</td>
<td>18.1 (4.06)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>19.2 (2.87)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>19.2 (2.35)&lt;sup&gt;A&lt;/sup&gt;</td>
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<td>PNS perp OrPo (mm)</td>
<td>17.8 (2.66)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>19.5 (2.40)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>20.8 (2.82)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.001&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>OrPoANS–PNS (°)</td>
<td>0.4 (4.37)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>−0.4 (3.35)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>−1.6 (2.66)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.149</td>
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<td>Mandible</td>
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<tr>
<td>SNB (°)</td>
<td>78.4 (4.30)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>81.0 (3.68)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>77.2 (3.14)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.002&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>OrGoGoGn (°)</td>
<td>23.1 (5.00)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>26.1 (4.36)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>24.7 (4.27)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.045&lt;sup&gt;*&lt;/sup&gt;</td>
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<td>ArtGoMe (°)</td>
<td>127.6 (5.86)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>130.7 (4.28)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>130.4 (3.56)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.050</td>
</tr>
<tr>
<td>Go–Gn (°)</td>
<td>96 (7.71)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>105 (5.45)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>104.7 (5.48)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>Facial convexity and maxillomandibular relation</td>
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<tr>
<td>NAP (°)</td>
<td>2.6 (5.94)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.5 (8.46)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>6.4 (4.59)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.012&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>ANB (°)</td>
<td>1.4 (2.90)&lt;sup&gt;AB&lt;/sup&gt;</td>
<td>0.1 (4.11)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>3.6 (1.98)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.002&lt;sup&gt;**&lt;/sup&gt;</td>
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<td>MMD</td>
<td>20.6 (4.32)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>25.2 (4.09)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>19.9 (3.65)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<tr>
<td>Facial heights</td>
<td></td>
<td></td>
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<tr>
<td>N–Me (mm)</td>
<td>97.7 (7.28)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>104.5 (5.76)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>108.3 (5.59)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
</tr>
<tr>
<td>N–ANS (mm)</td>
<td>42.2 (4.60)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>45.4 (2.93)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>47.9 (3.46)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
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<td>ANS–Me (mm)</td>
<td>57 (4.50)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>60.8 (4.41)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>67.1 (3.38)&lt;sup&gt;B&lt;/sup&gt;</td>
<td>0.000&lt;sup&gt;**&lt;/sup&gt;</td>
</tr>
<tr>
<td>S–Go (mm)</td>
<td>62.7 (5.90)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>64.8 (3.81)&lt;sup&gt;AB&lt;/sup&gt;</td>
<td>66.7 (4.71)&lt;sup&gt;h&lt;/sup&gt;</td>
<td>0.009&lt;sup&gt;**&lt;/sup&gt;</td>
</tr>
<tr>
<td>Ar–Go (mm)</td>
<td>38.2 (5.24)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>39 (2.74)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>38.2 (5.24)&lt;sup&gt;A&lt;/sup&gt;</td>
<td>0.446</td>
</tr>
</tbody>
</table>

SD, standard deviation

1One-way analysis of variance. Tukey's test: the means with different letters (A, B, and C) indicate statistically different and 'AB' not statistically different from 'A' and 'B'.

*P < 0.05; **P < 0.01.
Craniofacial anomaly is already present at birth and becomes with a homogeneous group of subjects of same gender, age, (1988; et al.) and Fischer-Brandies (1986), the shorter anterior cranial base in DS group. The children were radiographed. However, some children, even with perfect head position. Although the focus of this study was to assess the craniofacial morphology in prepubertal subjects with DS, some ethical considerations on orthopaedic/orthodontic treatment may be raised (Becking and Tuinzing, 1991; Silva et al., 1997). According to Desai and Flanagan (1999), mental deficiency itself is not always an obstacle for the treatment, but DS patients are not always aware of their malocclusion and facial deformity. Recent improvements in medical care are keeping DS subjects healthy for longer and socially integrated. Therefore, orthopaedic/orthodontic treatment may offer an aesthetic and functional improvement in a carefully selected group of patients and thereby enhance their social acceptance. The present investigation reinforces the need for further studies on this issue, including a multidisciplinary treatment approach.

Figure 3  Schematic superimposition of the mean cephalogram of Down syndrome (dotted line), maxillary deficiency (dashed line), and control (skeletal Class I, solid line) groups. The superposition was made in the Frankfurt horizontal plane centred on pterygomaxillary fissure.

|                | Mean (SD) Mean (SD) Mean (SD) |
|----------------|-------------------------------|-------------------------------|
| Facial heights |
| Ar – Go (mm)   | 38.2 (5.24) A                 | 39 (2.74) A                   | 38.2 (5.24) A |
| N – ANS (mm)   | 42.2 (4.60) A                 | 45.4 (2.93) B                 | 47.9 (3.46) B |
| ANB (°)        | 1.4 (2.90) AB                 | 0.1 (4.11) A                 | 3.6 (1.98) B  |
| Go – Gn (mm)   | 96 (7.71) A                   | 105 (5.45) B                 | 104.7 (5.48) B |
| OrPoGoGn (°)   | 23.1 (5.00) A                 | 26.1 (4.36) B                 | 24.7 (4.27) AB |
| SNB (°)        | 78.4 (4.30) A                 | 81.0 (3.68) B                 | 77.2 (3.14) A |
| ANS perp OrPo (mm) | 18.1 (4.06) A           | 19.2 (2.87) A                 | 19.2 (3.25) A |
| BaSN (°)       | 134.6 (4.90) A                | 126.4 (5.60) B                | 128.1 (2.85) B |
| ArGoMe (°)     | 127.6 (5.86) A                | 130.7 (4.28) A                | 130.4 (5.36) A |

A greater maxillary involvement in DS compounded a less convex facial profile (NAP), when compared with the control group, but not in relation to the maxillary deficiency group, which was more concave. The MMD relationship (ANB and MM) was intermediate in the DS group, with the worst relation observed in the maxillary deficiency group. This facial feature composition could lead to a mistaken concept about mandibular growth in the DS group. In the present study, DS presented a shorter body length (Co–Gn), although it was spatially well positioned with respect to the cranial base (SNB).

Reduction in the anterior facial heights (N–Me, N–ANS, and ANS–Me) in DS was not accompanied by posterior heights (Ar–Go and S–Go), which was similar to maxillary deficiency and control groups, supporting the typical short face (brachycephaly; Motohashi, 1985; Fischer-Brandies, 1988; Allanson et al., 1993). Suri et al. (2010) found a contrasting result in the posterior height in adolescent DS. This might probably be due to the younger age of the studied sample (8.3 years, ranging from 6.5–11.7 years), in which the differences had not been expressed fully.

Although the focus of this study was to assess the craniofacial morphology in prepubertal subjects with DS, the present study was the recruitment of a pure maxillary deficiency sample. Some children may have a mildly protrusive mandible associated with a maxillary deficiency, thus affecting comparisons. Furthermore, the clinical and cephalometric diagnosis in borderline cases causes a persistent doubt. The DS subjects in this study presented smaller mandibular length, confirming the characteristic of the syndrome. Likewise, mandible morphology (ArGoMe) was similar in both the groups in the same way.

degrees) when compared with the nonsyndromic groups (BaSN = 126.4 degrees and 128.1 degrees; Table 1). Furthermore, on comparison with the literature, DS of the present study was found to be similar (BaSN = 136.8 degrees) to another study (Fischer-Brandies et al., 1986) for the corresponding age. These cranial base changes for angular and linear measurements, which are very well documented in the literature (Frostad et al., 1971; Fink et al., 1975; Motohashi, 1985; Fischer-Brandies et al., 1986; Fischer-Brandies, 1988), suggest that antero-posterior and vertical cephalometric measurements for maxilla and mandible, based only on altered cranial base, can lead to an equivocal cephalometric interpretation and hence should be avoided. Hence, in the present study, measurements of the vertical position of the maxilla and mandible were based on the Frankfurt horizontal plane. This reference is considered more stable and accurate (Ellis and McNamara, 1988), although the palatal plane showed no expressive rotation in relation to the Frankfurt horizontal plane used in this study or anterior cranial base (S–N) from other studies (Fischer-Brandies et al., 1986; Fischer-Brandies, 1988). In addition, in the present study, cephalometric measurements that relied on the Frankfurt horizontal plane (ANS perp OrPo and PNS perp OrPo) showed high systematic error and low reproducibility.

The SNA angle showed no statistical difference among the three groups (Table 1). However, effective maxillary length (Co–A) was markedly shortened in the DS group and was statistically different among the other groups. The shorter hard palate had been considered as one of the typical signs in DS (Austin et al., 1969). In addition, Fischer-Brandies (1988) also evidenced greater maxillary involvement, with a 5-mm reduction in the palatal length.

Relative macroglossia refers to a large and hypotonic tongue relative to the bony confines of the oral cavity but with an absolute volume smaller than that of the normal tongue (Guimaraes et al., 2008). The reduction in facial dimensions with a large and hypotonic tongue may predispose DS subjects to a high prevalence of obstructive sleep apnoea (Guimaraes et al., 2008), and not surprisingly, some DS subjects are treated with maxillary expansion (Silva et al., 1997), surgical maxillary advancement (Janson et al., 2009), partial glossectomy (Wolford and Cottrell, 1996), and growth hormone therapy (Carlstedt et al., 1999).

Mandibular base length (Go–Gn) was similar between maxillary deficiency and control groups, with a greater projection in the maxillary deficiency group (SNB; Table 1). One difficulty in the present study was the recruitment of a pure maxillary deficiency sample. Some children may have a mildly protrusive mandible associated with a maxillary deficiency, thus affecting comparisons. Furthermore, the clinical and cephalometric diagnosis in borderline cases causes a persistent doubt. The DS subjects in this study presented smaller mandibular length, confirming the characteristic of the syndrome. Likewise, mandible morphology (ArGoMe) was similar in both the groups in the same way.
Conclusion

Based on a cephalometric analysis, the findings of this study show that prepubertal children with DS differ from those with maxillary deficiency of the same age, gender, and ethnicity, with respect to the cranial base, which was found to be flatter and shorter on its anterior site, along with the general reduction in linear maxillary and mandibular length, as well as anterior facial heights. Despite the highly compromised maxillary length, the MMD sagittal relationship was observed to be less deteriorated and the maxillary deficiency was not so expressive in the face of DS children because of the overall reduction of craniofacial dimensions.

References

Motohashi N 1985 Craniofacial dysmorphology in syndromes associated with abnormal physical growth. Journal of Craniofacial Genetics and Developmental Biology 1: 211–225