British Journal of Medicine & Medical Research
17(5): 1-7, 2016, Article no.BJMMR.28513
ISSN: 2231-0614, NLM ID: 101570965

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Cephalometric Characteristics of Down Syndrome in Brazilian Population

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Authors' contributions

This work was carried out in collaboration between all authors. Author TFS designed the study. Authors JMPLF, JDMM and SESM collected the data. Authors TFS, NCP and ADV analyzed the cephalometric data. Authors TFS, JMPLF and CCOS drafted the article. All authors read and approved the final manuscript.

Article Information

DOI: 10.9734/BJMMR/2016/28513
Editor(s):
(1) Paulo Ricardo Gazzola Zen,Departament of Clinical Medicine, Federal University of Health Sciences of Porto Alegre (UFCSPA), Brazil.
Reviewers:
(1) Mainul Haque, National Defence University of Malaysia, Malaysia.
(2) Kritika Jangid, Smile Way Dental Care and Research Centre, Rajasthan, India.
Complete Peer review History: http://www.sciencedomain.org/review-history/15808

Original Research Article

Received 22nd July 2016
Accepted 10th August 2016
Published 16th August 2016

ABSTRACT

Objective: This study aimed to compare cephalometric characteristics between individuals with Down syndrome (DS) and non-syndromic subjects.
Methods: Teleradiography of 15 patients with Down syndrome and 15 nonsyndromic individuals matched by age and gender were analyzed. Statistical analysis was performed comparing cephalometric measurements between groups using Mann-Whitney test.
Results: The analysis of measurements showed the anterior and posterior shortening of the cranial base (SN and SBa length) in individuals with DS ($p<0.05$). Regarding the skeletal sagittal plane, it was noticed a reduction of SNA and SNB angles in cases with DS ($p<0.05$). The parameters ANB, Co-A and Co-Gn of individuals with Down syndrome were significantly reduced

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when compared to non-syndromic individuals (p<0.05). The 1U-NA angle presented was increased in cases with DS (p=0.001).
Conclusions: Individuals with Down syndrome have shortening of anterior and posterior cranial base, maxilar and mandibular retrognathia and protrusion and proclination of upper incisors.

Keywords: Down syndrome; cephalometrics; craniofacial; dental.

1. INTRODUCTION

Down's Syndrome (DS), also known as trisomy 21 (OMIM #190685), is a chromosomal abnormality characterized by increased genetic material from chromosome 21 [1]. In most cases DS is characterized by the whole chromosomal aneuploidy. However, 5% occurs in the form of translocations and mosaics [2]. The literature describes that the only well-established risk factor for trisomy 21 is advanced maternal age (≥ 35 years at delivery), which is associated with an increased risk for trisomy 21 [3].

This syndrome was named for John Langdon Down in 1866, who described many of its features [4]. This condition is characterized by impairment in cognitive and motor development affecting between 1/600 to 1/2000 live births in the world [5]. The gender profile of DS showed typical male prevalence in subjects with regular trisomy 21 [6]. Apparently, the DS has no racial, socioeconomic or gender preference, though advanced maternal age may be associated with increased prevalence of this condition [1,5].

Regardless of ethnicity, DS has a combination of typical facial features such as reduced skull size, midface deficiency, nasal bone depression, straight facial profile, plus malocclusions mainly characterized by deviations in shape, size and/or position of the maxilla and mandible [1,7].

The cephalometric analyzes, carried out from the lateral radiograph of the face, has been shown to be a very useful tool for the characterization of the skull and face of individuals with DS [8]. Through cephalometry was possible to identify features such as: reduced overall size of the skull, jaw and mandibular hypoplasia, previous reduction of the cranial base and presence of open bite [9-13].

In Brazil, it is estimated that there are 300,000 individuals with Down syndrome [14]. However, there is a lack of studies that address its development and craniofacial characteristics in Brazilian DS individuals. In this sense, the aim of this study was to investigate the cephalometric characteristics of Brazilian individuals with DS.

2. MATERIALS AND METHODS

2.1 Study and Sample

This was a retrospective cross-sectional study performed with a sample of convenience on 30 Brazilian adults of both genders divided into two groups: (1) a group consisted of 15 patients with DS (case group); (2) a group consisted of 15 nonsyndromic patients (control group). All subjects in this study were recruited in the city of Juazeiro do Norte, state of Ceará, northeastern Brazil.

The groups were matched by gender and none of the subjects had past history of orthodontic treatment. All patients with Down`s syndrome were recruited from non-governmental institution. All individuals with DS were diagnosed in specialized medical services of that institution. This study was approved by the Research Ethics Committee, protocol number 1.166.198.

2.2 Cephalometry

Lateral face radiographs of all selected patients (teleradiography) were used to carry out the cephalometric analysis. Radiographs were selected from the files of the Imaging Center at the School of Dentistry of Centro Universitário Leão Sampaio - UNILEÃO. All radiographs were acquired using the device x-ray Dabi Atlante® Eagle type. The procedures for obtaining the teleradiographs were performed by the same technician with specific training in the face of lateral radiographs.

Cephalometric analyzes were performed by the same examiner, an orthodontics specialist (TFS). Conventional cephalometric tracings were performed on all radiographs. The standard used for the analysis followed the parameters described by Gandini-Junior et al. [15] and Korayem and Alkofide [8]. The angular and linear measurements, as well as their definitions are arranged in Table 1.

Table 1. References and cephalometric settings

References cephalometric	Definitions
Cranial base	
SN length mm	It is the distance between sella point and nasion. Represents the anterior cranial base length.
SBa length mm	It is the distance between sella and basion point. Represents the posterior cranial base length.
NSBa angle	Angle formed by the intersection of SN and SBa lines. Shows the posterior cranial base inclination.
Sagittal skeletal	
SNA angle	Angle formed by the intersection of SN e NA lines. Shows the anteroposterior position of the maxilla relative to the cranial base.
SNB angle	Angle formed by the intersection of SN and NB lines. Shows the anteroposterior position of the mandible relative to the anterior cranial base.
Maxillo-mandibular relationship	
ANB angle	Angle formed by the intersection of lines NA and NB. Shows the maxillo-mandibular relationship in the anteroposterior direction.
WIT's mm	It is difference between the projection distance from A-point perpendicularly to the functional occlusal plane and the projection of B-point perpendicularly to the functional occlusal plane. Shows the maxillo-mandibular relationship in the anteroposterior direction.
Co-A mm	It is the distance between A-point and Co point. Shows the effective length of the maxilla.
Co-Gn mm	It is the distance between the B-point and Co point. Shows the effective length of the mandible.
Vertical skeletal	
LAFH mm	The lower facial height (LAFH) is the distance between the ANS and Me point. Shows the effective length of the anterior facial height.
Facial axis	Angle formed by the intersection of Ba N with PtGn line. Shows the growth pattern of the face.
Dental	
UI-NA angle	Angle formed by the intersection of the long axis of the upper incisor with the NA line. Indicates the axial inclination of tooth relative to bone base.
UI-NA mm	Linear distance from most buccal point of upper incisor crown to line NA. Indicates the protrusion of the upper incisor.
LI-NB angle	Angle formed by the intersection of the long axis of the lower incisor with the NB line. Indicates the axial inclination of tooth relative to bone base.
LI-NB mm	Linear distance from most buccal point of lower incisor crown to line NB. Indicates the protrusion of the lower incisor.
UI-LI angle	Angle formed by the intersection of the long axes of the upper and lower incisors. Indicates the degree of protrusion of the incisors.
Soft tissue	
Nasolabial angle	Angle formed by Col-Sn-UL. Indicates the degree upper lip protrusion.

The cephalometric points used in this study are shown in Fig. 1. To ensure the reliability of cephalometric analysis, an intra-examiner test with 10 radiographs (05 DS and 05 nonsyndromic) was performed. Concordance analysis was more than 85% (Kappa > 0.85), reflecting a satisfactory agreement.

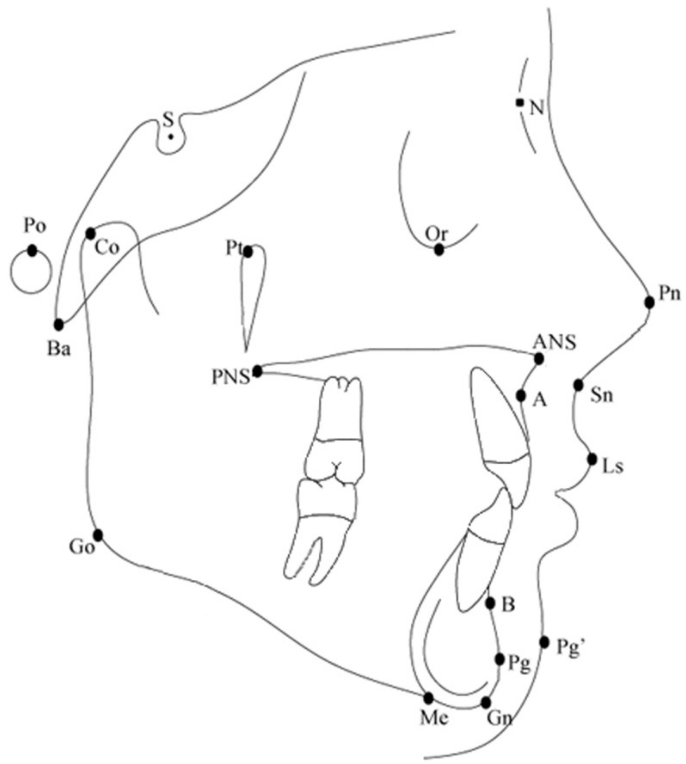

Fig. 1. Cephalometric points used in the study

Po indicates porion; S, sella; Or, orbita inferior; N, nasion; Ba, basion; Pt, pterygoid; B, supramentale; Pg, pogonion; Gn, gnathion; Me, menton; Go, gonion; A, subspinale; ANS, anterior nasal spine; Co, condylion; Sn, subnasale; Pn, pronasale; Ls, labia superioris; Pg', soft tissue pogonion; and PNS, posterior nasal spine

3. RESULTS

From the 30 patients selected for this study, 15 (50%) were male and 15 (50%) were female. Mean age was 27 years (median 29 years), ranging from 21 to 34 years old. The cephalometric analyzes of this study are shown in Table 2.

The analysis of cranial measurements showed anterior and posterior shortening of the cranial base (SN and SBa length) in individuals with DS ($p \leq 0.05$). In addition, the SN-Ba presented angle was increased in these patients ($p=0.011$), indicating an increased slope of the skull base when compared to nonsyndromic individuals.

Regarding skeletal measurements in the sagittal plane, it was noticed a reduction of SNA and SNB angles in cases with DS ($p \leq 0.05$). This finding highlights the maxillary and mandibular retraction in patients with DS. When analyzing the maxillomandibular relationship, ANB parameters, Co-A and Co-Gn of individuals with Down syndrome were significantly reduced when compared to nonsyndromic individuals ($p \leq 0.05$), showing a tendency to class III and reducing the size of maxilla and mandible.

The study of vertical magnitudes showed a brachyfacial trend in subjects with DS. The LAFH and facial axis were reduced in this group. However, it is noteworthy that this result was not statistically significant.

The comparison of dental patterns between groups showed no significant difference regarding the parameter analyzes: UI-NA measure, LI-NB angle, LI-NB measured and UI-LI angle. However, the UI-NA angle presented was increased in cases with DS ($p=0.001$) showing the axial inclination of the upper incisors.

4. DISCUSSION

Down syndrome is one of the most prevalent genetic conditions in the general population [8,16], characterized by varying degrees of

Table 2. Comparison of cephalometric measurements between groups

References cephalometric	Patients DS (n=15)		Nonsyndromic patients (n=15)		*P** value
	Mean rank	Mean	Mean rank	Mean	
Cranial base					
SN length mm	5.70*	63.0	15.30*	73.5	**0.000**
SBa length mm	5.50*	36.7	15.50*	52.2	**0.000**
NSBa angle	13.85*	151.5	7.15*	140.3	**0.011**
Sagittal skeletal					
SNA angle	5.50*	70.6	15.50*	83.9	**0.000**
SNB angle	5.50*	70.3	15.50*	83.9	**0.000**
Maxillomandibular relationship					
ANB angle	7.75*	2.0	13.25*	4.25	**0.036**
WIT's mm	12.1*	5.6	8.9*	4.6	0.220
Co-A mm	5.95*	73.8	15.05*	94.7	**0.001**
Co-Gn mm	6.10*	107.4	14.90*	135.3	**0.001**
Vertical skeletal					
LAFH mm	9.50*	65.3	11.50*	69.2	0.448
Facial axis	8.80*	89.7	12.20*	90.8	0.191
Dental					
UI-NA angle	14.80*	36.8	6.20*	24.2	**0.001**
UI-NA mm	11.45*	8.4	9.55*	6.87	0.468
LI-NB angle	9.20*	25.5	11.80*	28.2	0.325
LI-NB mm	8.10*	3.75	12.90*	6.7	0.068
UI-LI angle	8.25*	119.3	12.75*	125.5	0.088
Soft tissue					
Nasolabial angle	9.15*	98.4	11.85*	103.2	0.307

** Mann-Whitney test*

mental and physical disabilities [17]. In Dentistry, the knowledge of the craniofacial changes of individuals with DS is highly important for establishing the orthodontics diagnosis as well as the plan of treatment. At the craniofacial region, the literature describes that small nose, narrow deep and high palate, bifid uvula, underdeveloped jaw, cleft palate, inadequate lip closure, hypotonic lips, fissured tongue, inaccurate and slow movement of the tongue and changes in temporary and permanent tooth eruption are common findings to DS [18-20]. However, studies for the cephalometric characteristics of DS are still scarce, especially in Brazil.

The results of this study demonstrated significant differences in the cephalometric findings between groups. The first part of the analysis has focused on the measurement of the anterior length of the cranial base (SN), posterior length of the cranial base (SBa) and posterior angle of the cranial base (SNBa). The findings of this study showed that anterior and posterior length of the cranial base were reduced in DS individuals when compared with nonsyndromic individuals, suggesting a reduction of length of cranial base of these individuals. This fact may be related with steepening of the posterior cranial base. The literature describes that the cranial base angle is more obtuse and the length of the front and back of the cranial base presents shorter in DS subjects when compared with nonsyndromic individuals [8,21]. Fink et al. [9] concluded that the area of the endocranium demonstrated significantly lower in DS group and remained lower with advancing age.

The second part of the analysis was focused on the skeletal magnitudes of the maxilla and mandible. Our results showed a posterior positioning of the maxilla and mandible relative to the cranial base (SNA and SNB) in patients with DS, suggesting maxillomandibular retrognathia. In addition, Co-A and Co-Gn magnitudes showed a reduction in the length of the maxilla and mandible, which may establish a maxillary and mandibular micrognatia. The current findings regarding the size and maxillary and mandibular positioning in DS are scarce. Evidence supports that individuals with DS have reduced craniofacial complex and maxillary retrognathia, and show the average area of the face significantly lower than normal individuals [8-10,12,21,22].

The maxillomandibular sagittal relationship, assessed by the ANB angle, showed a skeletal

Class III tendency in the case group. In the study of Alió et al. [22], the researchers reported that people with Down syndrome have maxillary retrusion especially in the horizontal plane. Additionally, Quintanilla et al. [13] concluded that the mandibular position was shown within the normal range in the lower third of the face. These facts suggest the DS subjects have a tendency to skeletal Class III with maxillary deviation. However, it is noteworthy that our results showed a possible retrognathia and micrognathia mandibular, that, when associated with the size and positioning of the maxillary, may establish a retraction of the middle and lower third of face in individuals with SD, and can generate sagittal deviations in maxillomandibular positioning. Regarding the mandibular length, Korayem et al. [8] reported no significant difference between syndromic and nonsyndromic individuals. This difference in results may be linked to inappropriate position of mandible during the radiographic examination. Another important point is that individuals with DS may show macroglossia and hypotonic oral musculature. Thus, it is possible that the mandible of patients slid during the X-ray exam. Such a situation could lead to erroneous data about the mandibular positioning of these patients.

About the vertical magnitudes, our results suggest a tendency to brachyfacial facial type in DS group. The LAFH and the angle of the facial axis were reduced in the DS group compared with the control group, but was not statistically significant. The results of Quintanilla et al. [13] revealed that DS patients show a normal pattern of mandibular growth, rather than vertical growth. Korayem et al. [8] reported an increase in vertical relationship in patients with DS increased, which may lead to a tendency to open bite. These different results may be related to population and ethnic factors specific to the study populations.

The analysis of the dental patterns of DS individuals with the control group showed an increase in axial inclination of the upper incisors labially in the case group. Suri et al. [21] described that patients with DS have proclination of the upper incisors. Additionally, Korayem et al. [8] concluded that individuals with DS have the upper central incisors protruded and proclined, as well as reduced interincisal angle. The anterior tooth inclination suffers direct influence of the lip muscles and tongue. The macroglossia and labial hypotonia may be associated with increased dental anterior protrusion in patients with DS [1,8,14,21].

Regarding soft tissue analysis, the results showed a downward trend in the nasolabial angle in the case group. Korayem et al. [8] concluded that the nasolabial angle in DS patients are more acute than in nonsyndromic patients. These findings suggest that vestibule version of the upper incisors may reduce the nasolabial angle in individuals with DS. However, Sforza et al. [23] reported that the nasolabial angle can be increased in DS individuals.

Cephalometric differences observed in this study may help establish what the typical facial skeletal characteristics of individuals with DS. However, it is important to note that ethnic and racial characteristics of individuals may generate different results in different populations. This is one of the first cephalometric studies in individuals with DS in particular with a population of Northeastern Brazil. However, it is noteworthy that studies involving larger samples are needed to strengthen the typical cephalometric findings from Brazilian subjects with DS.

5. CONCLUSION

The presented anterior and posterior length of the cranial base is reduced and maxilla and mandible are retruded in individuals with DS. The present study has shown that the length of the maxilla and mandible is reduced and upper central incisor is protruded in individuals with DS.

CONSENT

All authors declare that written informed consent was obtained from the patients (or other approved parties) for this study.

ACKNOWLEDGEMENTS

The authors are grateful to the Associação de Pais e Amigos dos Excepcionais – APAE and the Centro Universitário Leão Sampaio - UNILEÃO for their cooperation with this study.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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Peer-review history:
The peer review history for this paper can be accessed here:
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