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Original Research Article

Evaluation of anthropometric facial landmarks in woman with Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome (BPES)

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Abstract

Introduction: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) is a syndrome easily recognized by facial appearance. In this sense, the facial anthropometry is a simple and non-invasive way to evaluate the morphology of the facial surface of individuals, thus, defining the craniofacial dimensions. **Objective:** To evaluate the facial anthropometric measurements of a Caucasian female, aged 20 years, diagnosed with BPES and to compare these measures with the values described in the literature for non-syndromic woman. **Material and methods:** This research is an observational study of a Caucasian female, aged 20 years, who was diagnosed with BPES. Frontal photographs were taken, and the images analyzed by nine researchers calibrated in Image J® software. The facial measurements evaluated were head, face, orbits, nose, and labio-oral region and were compared with non-syndromic woman. **Results:** All vertical and horizontal face measurements were higher than that of other females from Caucasian groups. BPES woman also presented bilateral ptosis and the main differences appear in the region of the orbits. **Conclusion:** The anthropometric facial analysis of BPES woman showed a significant change in the facial landmarks.

Introduction

Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) has been described in the literature at the beginning of the 20th century and affects mainly the eye attachments [3]. The main clinical features of all types are facial phenotypic appearance characterized by shortening of the horizontal distance from the orbital fissure associated with dysplasia of the upper eyelid muscle, which causes drooping of the upper eyelid or ptosis, and inversion of the lower eyelid skin in the medial region of the eyes [11]. Raised arched eyebrows and strabismus are also observed [5]. To compensate for the small eye opening, people with BPES take on a characteristic posture with the head tilted back and frowning [8].

Regarding the recognition of the facial features of syndromes, there is some difficulty since each syndrome has characteristics that identify them, but errors are not rare due to the similarity between the different syndromes. In this sense, the facial anthropometry helps in taking action from standardized physical landmarks of the face of the person and allows, in a simple and non-invasive way, to evaluate the morphology of the facial surface of individuals, thus, defining the craniofacial dimensions in different syndromes [2].

Standardized physical landmarks are crucial to reduce measurement error and increase reliability in anthropometry. The successful prior use of craniometric landmarks makes extrapolation of these landmarks to the soft tissue and facial photogrammetry [4], and can be used to determine the pattern of facial growth, predicting probable dysfunctions in craniofacial structures, dental occlusion, and the orofacial muscles [10].

Therefore, the goal of these study was to evaluate the facial anthropometric measurements of a Caucasian female, aged 20 years, diagnosed with BPES and to compare these measures with the values described in the literature for non-syndromic females.

Material and methods

This research is an observational study of a Caucasian female, aged 20 years, who had been diagnosed with BPES and was assisted in the clinic of Dentistry for Patients with Special Needs at PUCPR. Data about medical and dental history was obtained. After the signing of the consent informed by the mother, frontal photographs were taken. Therefore, the woman was positioned sitting with her head up, looking at the horizon. Cross imaginary plane passing between the eyes was parallel to the ground and the lens of Sony® camera was positioned 1.0 meter away from the face of the woman and the image was taken by positioning a millimeter ruler next to the head.

The images were analyzed by nine researchers calibrated in Image J® software by which the facial measurements were evaluated (table I, figure1).

Table I – Landmarks used to facial anthropometric measurements

Measurement	Landmark
Lower facial width	Gonion to gonion (go-go)
Mouth width	Cheilion to cheilion (ch-ch)
Nasal width	Alare to alare (al-al)
Upper facial width	Zygion to zygion (zy-zy)
Outer canthal distance	Exocanthion to exocanthion (ex-ex)
Intercanthal width	Endocanthion to endocanthion (en-en)
Eye fissure length	Exocanthion to endocanthion (ex-en/right eye)
Eye fissure length	Endocanthion to exocanthion (en-ex/left eye)
Intercanthal width	Endocanthion to endocanthion (en-en)
Forehead height	Trichion to nasion (tr-n)
Physiognomic face height	Trichion to gnathion (tr-gn)
Morphological face height	Nasion to gnathion (n-gn)
Lower face height	Subnasale to gnathion (sn-gn)

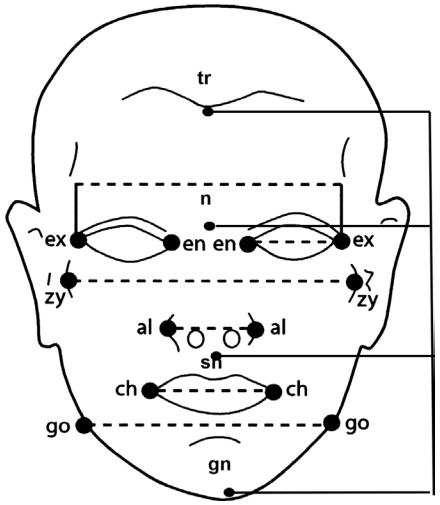

Figure 1 – Measurements on the frontal aspect of the face: Orbits: en-en (intercanthal width) ex-ex (biocular width) en-ex (eye fissure length) Face: zy-zy (face width) go-go (mandible width) Nose: al-al (morphological nose width) Labio-oral region: ch-ch (mouth width). Head: tr-n (forehead height) Face: tr-gn (physiognomic face height) n-gn (morphological face height) sn-gn (lower face height)
Source: Adapted from Farkas *et al.* [6]

Calibrated researchers performed all measurements and the obtained anthropometric values were compared with profile pattern studies involving the population of Caucasian women in the same age group.

Results

The facial measurements of the woman with BPES are summarized in table II. Regarding the vertical face measurements (tr-gn, sn-gn, n-gn, tr-gn), all of them were higher than those of the other females from Caucasian groups, but the two lower third of the face were significantly bigger (n-gn = 149.5 mm). Also, the physiognomic face height (tr-gn) was greater than that of the other groups.

Table II – Anthropometric measurements from the woman with BPES compared with other Caucasian women. The woman has the same age range of Caucasian women

Measurements	BPES Female	German Females	Hungarian Females	Italian Females	Polish Females	Portuguese Females
al-al	41.8	31.0	33.5	29.5	32.6	31.9
go-go	132.3	91.5	95.0	104.9	93.9	84.3
zy-zy	132.5	123.4	131.3	133.3	135.5	120.4
ch-ch	59.1	48.2	51.6	47.7	49.0	45.3
ex-ex	105.2	86.4	97.3	89.5	87.4	93.9
ex-en (right eye)	29.7	31.8	39.7	32.7	32.8	35.9
en-ex (left eye)	31.7	31.8	39.7	32.7	32.8	35.9
en-en	41.6	28.6	31.2	27.6	29.2	29.1
tr-n	78.7	66.3	69.2	59.4	63.7	62.2
sn-gn	79.5	63.3	56.7	64.4	60.5	62.8
n-gn	149.5	109.5	112.4	113.8	111.6	118.2
tr-gn	228.2	170.9	169.4	171.4	172.1	177.4

* All measurements were taken in millimeters (mm). Data referring to the other Caucasian women were obtained in Farkas *et al.* [6]

The horizontal face measurements revealed a large mandible width (go-go =132.3 mm), greater width of the nose (al-al = 41.8 mm) and mouth width (ch-ch = 59.1mm), and a similar distance between zygomatics (zy-zy= 132.5 mm).

Morphologically, the woman with BPES presented bilateral ptosis, so the main differences appear in the region of the orbits: intercanthal width (en-en), binocular width (ex-ex), and eye fissure length (en-ex or ex-en) differ widely, including orbital fissure of the right eye that differs from the orbital fissure of the left eye.

Discussion

The clinical diagnosis of a syndrome, especially given their diversity, is difficult. This has led some anatomists to explore physical landmarks to objectively assess patients [1]. Craniofacial anthropometry provides a simple and non-invasive method of quantitative assessment of changes on the surface anatomy of the head and face in individuals with a syndrome [2]. It is an easy non-invasive method of diagnosis. The goal of this observational study was to evaluate the facial features in a Caucasian woman with BPES and compare the results with morphometric data of the non-syndromic population. The woman analyzed in this study has European ascending so, for comparison, we used the study of Farkas *et al.* [6] that analyzed European Caucasian groups.

It is known that facial growth occurs in the vertical and horizontal direction and that there are three basic facial forms: long face or dolichofacial, medium face or mesofacial, and short face or braquifacial. Despite small differences between European groups, craniofacial analysis of the woman with BPES showed differences in vertical and horizontal measurements. The vertical measurements alone could point out to a dolichofacial aspect, but the horizontal measurements are greater too, so the female with BPES presents a larger face than that of women in her same age group.

As indicated above, anthropometric landmarks are often used jointly in craniofacial analysis and the vertical face measurements (tr-gn, sn-gn, n-gn, tr-gn) showed a physiognomic face height greater than that of other Caucasian groups, but the two lower thirds of the face were significantly wider (n-gn = 149.5 mm). This finding is confirmed by measuring the width of the mandible (go-go =132.3 mm), of the nose (al-al = 41.8 mm), and mouth (ch-ch = 59.1 mm), all significantly higher than Caucasian groups. Alterations in the morphology of the mandible may occur due to the positioning of the tongue [7]. This situation was observed in the woman with BPES.

The middle and upper thirds of the face reveal different morphometric findings: the distance between zygomatics is the measure that is closest to non-syndromic women (zy-zy = 132.5 mm), but the measurements of the orbits region differ greatly showing the main craniofacial aspect characterized by a shortening of the horizontal distance from the orbital fissure. These results were confirmed in this study: biocular width (ex-ex = 105.2 mm) measurement plus intercanthal width (en-en = 41.6 mm) were significantly larger than that of Caucasian females. The third measure for the orbits is the eye fissure length in relation to German females, but there is a difference between eyes (en-ex, left eye = 31.7 mm; ex-en right eye = 29.7 mm). These three horizontal measurements for the orbits that are exposed to visual judgment [6] and in case of the BPES, is frequently a pronounced reduction in the vertical angle of vision and contribute to facial features in BEPS patients.

The data obtained from the anthropometric analysis of the woman reveals that her face is greater in the lower two-thirds, and the upper third resembles that of the non-syndromic women, giving an aspect of a trapezoidal geometry with larger base facing down.

This study is a single description of craniofacial aspects of a female with BEPS. We know that factors like environmental conditions, socioeconomic status, physiological functions or disfunctions could contribute to variations in facial morphology, but, in case of syndrome, there appears to be a face pattern, so the description of this standard can help the dental planning for each case.

Conclusion

Despite this study limitations, the anthropometric facial analysis of woman with BPES showed a significant change especially in the lower third of the face and in the region of the orbits.

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