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CRANIOFACIAL DISPROPORTIONS IN WAARDENBURG SYNDROME TYPE I

Elias Oliveira da-Silva[1,2] and José Edmilson Mazza Batista[3]

ABSTRACT

Based on a set of 15 surface measurements, taken directly from the head and face of 51 patients with Waardenburg syndrome type I (WSI), values for 15 proportion indices were determined. Pairs between patients and controls of the same sex and age were formed for comparison. The main craniofacial disproportions observed in the WSI group were: wide inner intercanthal distance and narrow nose in relation to the outer intercanthal distance, and elongated ears and short philtrum compared with face height.

INTRODUCTION

The major clinical manifestations of Waardenburg syndrome type I (WSI) are telecanthus (*dystopia canthorum* or *dystopia canthi medialis*) with lateral displacement of lower lacrimal puncta, prominent nasal root, bushy eyebrows with synophrys, heterochromia or hypoisochromia iridis, hypopigmented ocular fundus, white forelock, premature greying, hypopigmented skin lesions and sensorineural deafness. A clinical and genetic study of WSI in two large Brazilian kindreds was previously described (da-Silva, 1991), having a gene mutation been recently reported in patients belonging to one of these families (Baldwin *et al.*, 1992).

The results of a clinical anthropometric and roentgencephalometric investigation, performed with most patients of both kindreds, were reported elsewhere (da-Silva *et al.*, 1993). Here, we analyze a variety of proportions in the craniofacial region of WSI patients based on a study of several indices, reflecting the relationships of surface measurements ot each other.

SUBJECTS AND METHODS

Based on a set of 15 surface measurements, values for 15 proportion indices (smaller measurement X 100/larger measurement) were determined. The measurements were taken directly from the head and face of 51 WSI patients (27 females and 24 males), ranging in age from four months of 71 years, and of an equal number of normal individuals. Pairs between patients and controls of the same sex and age (approximately) were established for comparison. The socioeconomic status and visible ethnic characteristics were also considered for the formation of the pairs.

The reference points (Figure 1) for taking the measurements were: alare (Al, the most lateral point at the alae nasi), cheilion (Ch, the lateral angle of mouth), endocanthion (En, the inner ocular canthus), euryon (Eu, the most lateral point at the parietal surface), exocanthion (Ex, the outer ocular canthus), glabella (G, the most anterior point between the supraorbital ridges), gnathion (Gn, the most inferior point on the mandibular symphysis), labrale superius (LS, the margin of the vermilion of upper lip), nasion (N, the most anterior point on the nasal root), opisthocranion (Op, the most posterior point on the occipital bone), postaurale (Pa, the most posterior point at the ear helix), preaurale (Pra, the most anterior point at the tragus), subaurale (Sba, the most inferior point at the ear

[1] Departamento de Genética, Universidade Federal de Pernambuco, Recife, PE, Brasil.

[2] Serviço de Genética Médica, Instituto Materno-Infantil de Pernambuco (IMIP), Rua dos Coelhos 300, Boa Vista, 50070-550 Recife, PE, Brasil. Send correspondence to E.O.S.

[3] Departamento de Estatística, Universidade Federal de Pernambuco, Recife, PE, Brasil.

lobe), subnasale (Sn, the point at which the columella merges with the upper lip), superaurale (Sa, the most superior point at the ear helix) and zygion (Zy, the most lateral point at the zygomatic arch). The following measurements were taken: occipitofrontal head circumference (OFHC), head length (G-Op), head width (Eu-Eu), morphological face height (N-Gn), face width (Zy-Zy), inner intercanthal distance (En-En), outer intercanthal distance (Ex-Ex), nose length (N-Sn), nose width (Al-Al), philtrum length (Sn-LS), mouth width (Ch-Ch), and length (Sa-Sba) and width (Pra-Pa) of both ears. Five of the indices expressed relations between measurements from the same anatomical region: cephalic (Eu-Eu X 100/G-Op), facial (N-Gn X 100/Zy-Zy), intercanthal (En-En X 100/Ex-Ex), nasal (Al-Al X 100/N-Sn) and auricular (Pra-Pa X 100/Sa/Sba) indices. The remaining 10 indices were interregional: face-head width index (Zy-Zy X 100/Eu-Eu), intercanthal-face width index (En-En X 100/Zy-Zy), biocular-face width index (Ex-Ex X 100/Zy-Zy), nose length-face width index (N-Sn X 100/Zy-Zy), nasofacial index (N-Sn X 100/N-Gn), nasozygomatic index (Al-Al X 100/Zy-Zy), nose-biocular width index (Al-Al X 100/Ex-Ex), philtrum-face height index (Sn-LS X 100/N-Gn), cheilozygomatic index (Ch-Ch X 100/Zy-Zy) and ear-face height index (Sa-Sba X 100/N-Gn).

A multivariate statistical analysis (stepwise and canonical discriminant analyses) was carried out in order to identify which of those indices best characterizes the syndrome. Pearson correlation coefficients (r) were calculated to compare the degree of relationship between the indices. The analyses were performed using Statistical Analyses System Software (SAS Institute Inc., 1987).

RESULTS AND DISCUSSION

Table I shows the descriptive statistical data. High correlations were only found between the following pairs of indices: facial/nose length-face width (r = 0.667), intercanthal/intercanthal-face width (r = 0.937), intercanthal-face width/nose-biocular width (r = 0.609), nose length-face width/nasofacial (r = 0.763) and nasozygomatic/nose-biocular width (r = 0.905).

In the stepwise discriminant analysis, five of the 15 indices were retained: intercanthal, face-head width, nose-biocular width, philtrum-face height and ear-face height indices. With the exception of the face-head width index, the other indices had significant F-tests at the 0.05 level. Thus, the main craniofacial disproportions observed in the WSI group were: wide inner intercanthal distance and narrow nose in relation to the outer intercanthal distance (increased intercanthal index and reduced nose-biocular width index, respectively), and elongated ears and short philtrum in relation to the face height

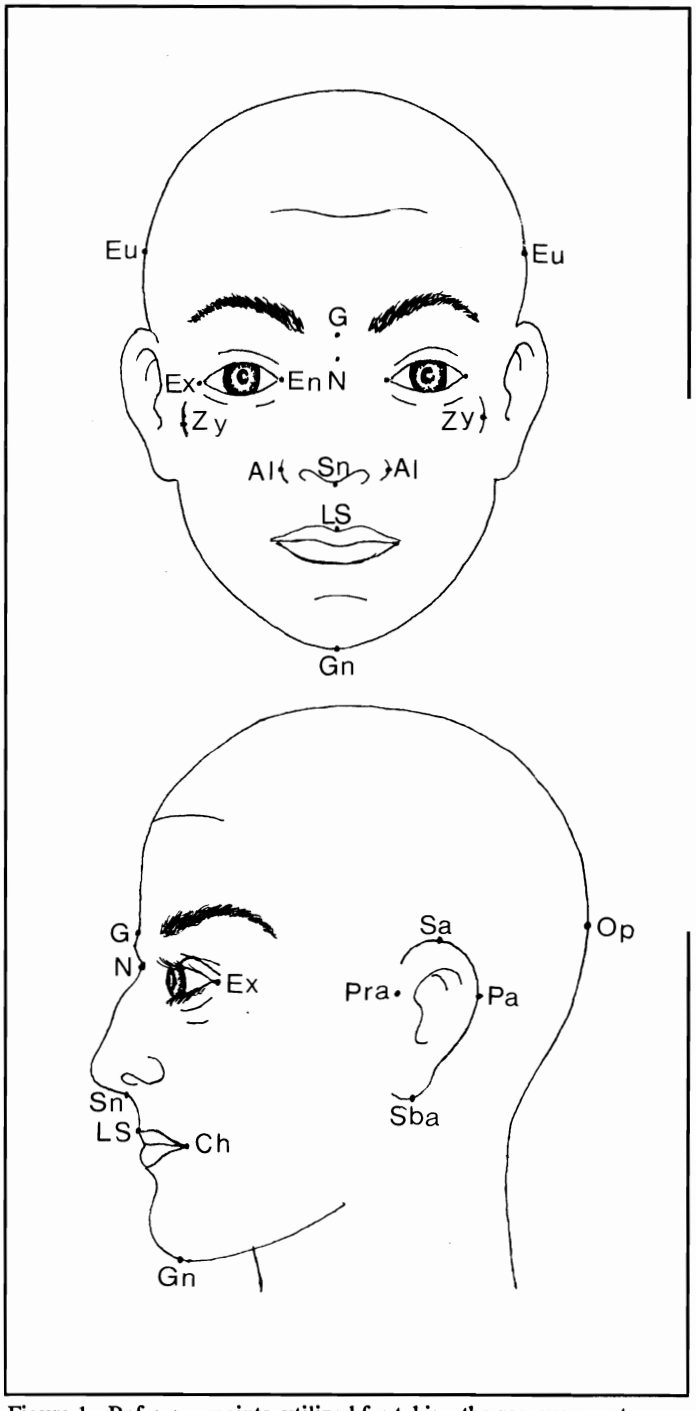

Figure 1 - Reference points, utilized for taking the measurements.

(increased ear-face height index and reduced philtrum-face height index respectively). An extensive analysis of craniofacial proportions was carried out by Farkas *et al.* (1985) in 28 cases of Apert syndrome (twenty females and eight males, with ages varying from 18 days to 15 years). Twenty indices, involving a total of 25 surface measurements, were investigated. The most frequent disproportions described in this syndrome were: wide inner intercanthal distance compared with nose width, brachycephaly, high head in relation to the face, narrow lower face in a relatively wide face, narrow nose compared

Table I - Descriptive statistical data: minimum (Min.), maximum (Max.), Mean (M) and standard deviation (SD).

Index	Group*	Min.	Max.	M	SD
Cephalic	1	68.42	96.97	82.92	5.27
	2	73.68	93.75	82.15	4.44
Facial	1	70.00	98.75	87.76	6.71
	2	73.74	99.36	88.33	6.12
Intercanthal	1	38.98	52.02	45.71	2.70
	2	30.14	38.64	34.76	1.91
Nasal	1	51.92	84.56	65.49	7.24
	2	51.30	95.35	72.75	8.24
Auricular	1	46.95	75.36	58.89	5.92
	2	46.10	68.35	56.97	5.55
Face-head width	1	72.79	92.34	83.05	4.73
	2	74.56	93.93	83.17	5.11
Intercanthal-face width	1	29.36	41.31	33.98	2.62
	2	20.69	29.90	25.33	1.90
Biocular-face width	1	65.05	83.13	74.38	4.60
	2	64.39	72.41	72.85	3.50
Nose length-face width	1	28.34	49.77	37.51	4.56
	2	28.38	47.81	38.47	4.19
Nasofacial	1	34.38	42.86	42.70	3.53
	2	33.68	53.87	43.59	3.99
Nasozygomatic	1	20.87	29.46	24.32	1.95
	2	21.36	36.10	27.78	3.03
Nose-biocular width	1	25.36	32.22	32.79	3.03
	2	29.10	48.99	38.19	4.36
Philtrum-face height	1	8.17	12.89	12.66	1.46
	2	11.30	20.41	15.63	2.09
Cheilozygomatic	1	28.52	49.40	37.12	4.00
	2	30.25	43.04	36.46	2.57
Ear-face height	1	47.89	64.50	54.51	4.14
	2	40.40	63.20	52.96	5.20

*1, Waardenburg I syndrome group; 2, normal control group.

to the face width and short mouth width in relation to the face width.

In our previous anthropometric/roentgencephalometric research with WSI (da-Silva *et al.*, 1993), the syndrome group had in general smaller craniofacies than normal individuals, and the most diagnostic parameters were: inner intercanthal distance, philtrum length, nasal bone length and lower facial height. In this study, the canonical discriminant analysis using the four significant indices showed a squared canonical correlation (R^2) of 0.891. Raw and standardized canonical coefficients are presented in Table II. These indices, as well as those physical parameters, had a high discriminating power for WSI, providing a 100% correct classification of patients and controls.

Table II - Canonical coefficients related to the most discriminating indices.

	Raw	Standardized
Intercanthal index	0.407	2.432
Nose-biocular width index	-0.087	-0.403
Philtrum-face height index	-0.202	-0.472
Ear-face height index	0.083	0.396

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RESUMO

Com base em um conjunto de 15 medidas de superfície, tomadas diretamente da cabeça e face de 51 pacientes com a síndrome de Waardenburg tipo I, foram determinados valores para 15 índices de proporção. Formaram-se pares, para comparação, entre pacientes e controles do mesmo sexo e idade. Foram as seguintes as principais desproporções observadas no grupo afetado: distância intercantal interna aumentada e nariz estreito em relaço à distância intercantal externa, e orelhas alongadas e filtro curto comparados com a altura da face.

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