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Photoanthropometric Investigation of Facial Structures in Iranian Children with Down Syndrome and Normal Controls

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ABSTRACT

The photoanthropometric method was used to study the facial features in 136 Iranian children with Down syndrome, aged 4 to 14 years. Nineteen parameters were investigated and compared to an age related control group of 100 normal Iranian children. The obtained measurements were related to reference values in the same faces. The normal range was defined by age related index values between the 20th and 80th percentile in the collective of normal Iranian children. Five parameters were considered as characteristic facial traits of Iranian children with Down syndrome by index values outside these percentiles in ≥ 50% of the studied collective: low midface; narrow and upslanted palpebral fissures ; short, and anteriorly rotated ears. Twelve parameters were considered as additional facial traits by index values outside these percentiles in ≥ 30% < 50% of the studied collective: broad inner canthal distance; prominent nose root; short nose back; everted nasal base; long nasolabial distance; forwards inclined integumental upper lip; narrow mouth fissure; high and prominent chin; high-set, narrow ears and narrow conchae. These results contribute to an objective definition of facial traits in children with Down syndrome in a homogeneous ethnic population.

INTRODUCTION

For the objective definition of dysmorphic traits in children with genetic syndromes, measurements of facial structures may be performed on photographs. Using the method of Stengel-Rutkowski et al. (1984), a total of 97 Caucasian children with different genetic syndromes were investigated using a cohort of 100 normal German children as controls: fragile X syndrome (n = 31), Prader-Willi syndrome (n = 37) and Williams-Beuren-syndrome (n = 29) (2,3,6). Here we present a similar investigation of facial structures in 136 Iranian children with Down syndrome, using a cohort of 100 normal Iranian children as controls.

MATERIAL AND METHODS

The 136 Tehranian children with cytogenetically proven Down syndrome and 100 normal Tehranian children were aged between 4 and 14 years. The normal children were picked up randomly at the cinema, swimming pool, bus station, etc. The only criterion for their inclusion in the normal collective was that they were not visibly mentally or physically handicaped. Facial photographs were taken in strict frontal and side view. The measuring points in the eye, nose, mouth and ear region were taken from the paper of Stengel-Rutkowski et al. (1984). The definitions of chin height, ear position and ear insertion line were modified and the inclination of palpebral fissures was newly defined.

The indices and angles were drawn as ordinates in a coordinate system with the age of the children as abscissa. The measuring points in the control group generally clustered within a well defined range. To check their distribution for age effects, the linear regression was calculated from the normal collective. Age dependency of facial parameters was defined by a correlation coefficient of r > 0.1. For the definition of the normal range, paralles were drawn to the regression line, separating 20% of the uppermost and the lowermost values, corresponding to the 20th and 80th percentiles.

For testing the accuracy, we repeated measurements from 10 frontal and profile photos of one 9 years old child. The standard deviation of the obtained indices varied between 0,31 and 2,94 representing a mean variance of 5.92% (Table 1).

RESULTS

In Figures 1 – 3 the index values of the facial parameters from the Iranian children with Down syndrome were drawn into the corresponding normal value diagrams elaborated on the basis of the index values from the collective of normal Iranian children.

Midface Height

75% of the children with Down syndrome had a low midface, defined by index values below the 20th percentile, 3% a high midface with index values above the 80th percentile. A moderate age effect was found in the normal collective (r = 0.33), indicating a slightly increasing midface height related to the bizygion diameter with increasing age (Fig. 1).

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Fig. 1. **Midface height (ophryon-stomion), related to bizygion diameter**

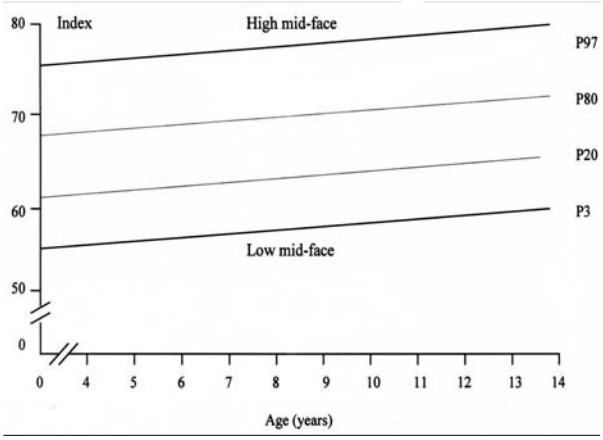
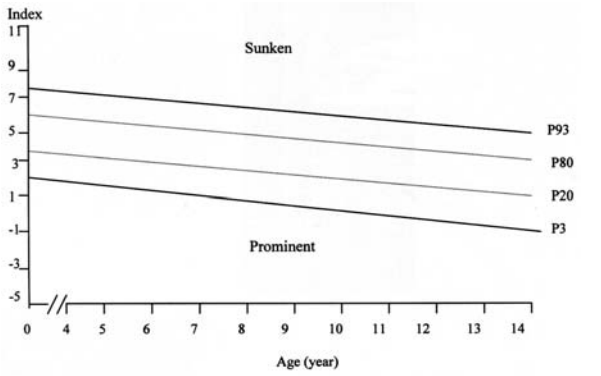


Fig. 2. Depth of the nose root (distance between profile line ophryon - subnasale and deepest point of the nose root), related to the midface height ophryon-stomion



Inner Canthal Distance
45% of the children with Down had a broad inner canthal distance (> p 80), 2% a narrow inner canthal distance (< p 20). A moderate age effect was found in the normal collective (r=-0.12), indicating a slightly decreasing inner canthal distance related to the bizygion diameter with increasing age.

Width of Palpebral Fissures
60% of the children with Down syndrome had narrow palpebral fissures (< p 20), 5% broad palpebral fissures (> p 80). A moderate age effect was found in the normal collective (r = 0.17), indicating a slightly increasing width of the palpebral fissures related to the bizygion diameter with increasing age.

Inclination of Palpebral Fissures
The inclination of the palpebral fissures, which was not measured in the previous study(8), was defined here by the angle between the palpebral fissure lines through the inner and the outer canthus of the left and the right eye on frontal photographs. 81% of the children with Down syndrome had laterally upslanting palpebral fissures (> p 80), 4% laterally downslanting palpebral fissures (< p 20). No age effect was found in the normal collective (r = - 0.06).

Depth of the Nose Root
34% of the children with Down syndrome had prominent nose roots (< p 20), 16% sunken nose roots. A distinct age effect was found in the normal collective (r=-0.54), indicating an uprighting nose root with increasing age (Fig. 2).

Length of the Nose Back
34% of the children with Down syndrome had a short nose back (> p 80), 29 % a long nose back (< p 20). A distinct age effect was found in the normal collective (r = 0.60), indicating an increasing nose lenght related to the midface heigth with increasing age.

Inclination of the Nasal Base
35% of the children with Down syndrome had an everted nasal base (< p 20), 8 % an inverted nasal base (> p 80). A distinct age effect was found in the normal collective (r = 0.45), indicating an increasing inversion of the nasal base related to the profile line with increasing age.

Interalar Distance
27% of the children with Down syndrome had a broad interalar distance (> p 80), 7% a narrow interalar distance (< p 20). A moderate age effect was found in the normal collective (r = 0.23), indicating a slightly increasing interalar distance related to the bizygion diameter with increasing age.

Prominence of the Maxillary Region
26% of the children with Down syndrome had a prominent maxillary region (> p 80), 24% a receding maxillary region. A distinct age effect was found in the normal collective (r = 0.42), indicating an increasing prominence of the maxillary region related to the subnasal radius with increasing age.

Nasolabial Distance
31% of the children with Down syndrome had a long nasolabial distance (> p 80), 10% a short nasolabial distance (< p 20). A moderate age effect was found in the normal collective (r = -0.30), indicating a slightly decreasing nasolabial distance with increasing age.

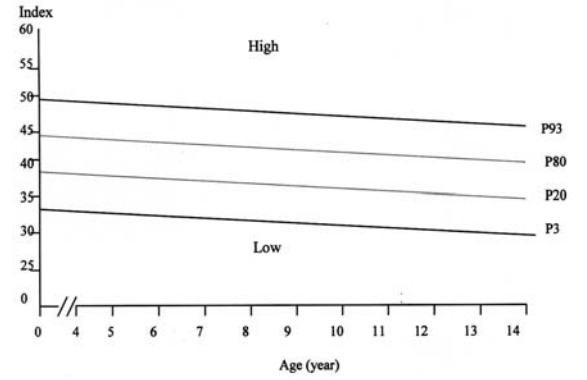
Inclination of the Integumental Upper Lip
35% of the children with Down syndrome had a forwards inclined integumental upper lip (procheilia; >p 80), 7% a backwards inclined integumental upper lip (retrocheilia; < p 20). No age effect was found in the normal collective (r = 0.001).

Mouth Width
32% of the children with Down syndrome had a narrow mouth fissure (< p 20), 11 % had a broad mouth fissure (> p 80). A moderate age effect was found in the normal collective (r = 0.26), indicating a slightly increasing mouth fissure with increasing age.

Chin Height
The index of Stengel-Rutkowski et al. (1984) was modified by using as reference the midface height instead of the total face height. The reason for this was the frequently open mouth in children with Down syndrome, which leads to an increase of the total face height, thereby relatively decreasing the index values

for the chin height. 47% of the children with Down syndrome had a high chin (> p 80), 15% had a low chin (< p 20). No age effect was found in the normal collective (r = - 0.01) (Fig. 3).

Fig. 3. **Height of the chin (distance between gnathion and tragion-sulcus mentolabialis line), related to midface height ophryon-stomion**



Chin Prominence
38% of the children with Down syndrome had a prominent chin (> p 80), 21% had a receding chin (< p 20). A distinct age effect was found in the normal collective (r=0.48), indicating an increasing chin prominence with increasing age.

Ear Position
The index definition of Stengel-Rutkowski et al. (1984) was modified by measuring the distance from the ophyron - instead from the outer canthus - to the horizontal to the profile line through the tragion. The reason for this was the laterally upturned eye axis in children with Down syndrome, which would lead to an increased frequency of low set ears. After the modified index definition, 30% of the children with Down syndrome had high set ears (p < 20), 26% had low set ears (p > 80). A distinct age effect was found in the normal collective (r = - 0.42), indicating an increasing ear position with increasing age.

Ear Inclination
Because of the laterally upturned eye axis, a modification was also made for this index definition by taking the subnasal radius instead of the tragion - outer canthus line (Stengel-Rutkowski et al. 1984) for the measurement of the angle to the otobasion superius - inferius line. 68% of the children with Down syndrome had anteriorly rotated ears (< p 20), 3% posteriorly rotated ears (> p 80). A moderate age effect was found in the normal collective (r = 0.12), indicating a slightly increasing posterior rotation of the ears with increasing age.

Ear Length
62% of the children with Down syndrome had short ears (< p 20), 4% long ears (> p 80). A moderate age effect was found in the normal collective (r = - 0.23), indicating a slightly decreasing ear length with increasing age.

Ear Width
47% of the children with Down syndrome had narrow ears (< p 20), 10% broad ears (> p 80). A moderate age effect was found in the normal collective (r = - 0.27), indicating a slightly reduced ear width with increasing age.

Concha Width
33% of the children with Down syndrome had a narrow concha (< p 20), 14% a broad concha (> p 80). A moderate age effect was found in the normal collective (r = - 0.23), indicating a slightly decreasing concha width with increasing age.
The results of the photoanthropometric measurements of these 19 facial parameters in the collective of 136 Iranian children with Down syndrome compared with the 100 normal Iranian children are summarized in Table 2.

DISCUSSION

The Photoanthropometric Method
The photoanthropometric method was used to determine objectively, whether a facial trait is normal or dysmorphic in the Down syndrome collective. Based on the definitions of measuring points, the investigated diameters were related to reference diameters within the same face and the obtained indices were compared to age related percentiles, which had been elaborated from the normal collective. This method is reproducible and constitutes an approach to quantitative dysmorphology. The needed instruments are readily available and the measurements easily performed. By modification of measuring points, three previous facial trait definitions were adapted to the pecularities of Down syndrome (chin height, ear position and insertion line) and one new parameter (eye axis) was introduced. The evaluation of measurements from children with Down syndrome and normal children from the same ethnic background eliminated the population specific variations.
Differences in mimics and head position during photography may lead to variations affecting the results. This was largest for the nasolabial distance and smallest for the inclication of the nasal base (Table 1). The mean variance was slightly below that obtained in the study in Germany in 1984, indicating a somewhat higher measuring accuracy(8). As all measurements were performed by the same person, there was no interobserver variability in this study.

The Iranian Normal Collective
To compare the Iranian and the German normal collective, the age effects were classified into three categories according to the obtained r values: no age effect (r < 0.1), moderate age effect (r = 0.1 - 0.4) and distinct age effect (r > 0.4). Differences were found for the following five facial parameters: 1) Inclination of the integumental upper lip: the Iranian data showed no age effect, while a moderate decrease of this angle was found with increasing age in the German data. 2) Inner canthal distance (slightly decreasing), 3) nasolabial distance (slightly decreasing), 4) mouth width (slightly increasing): moderate age effects were found in the Iranian data, while the German data showed no age effects. 5) Inclination of the nasal base: a distinct increase of this angle was found with increasing age in the Iranian data, while the German data showed only a moderate age effect.
Regarding the heights of the comparable indices and angles (intersection points of the regression lines on the vertical axis in the diagrams), 1/3 of the facial parameters differed between both normal collectives for ≥ 5 point values or degrees.

These results point to constitutional differences between Iranian and German normal children. Therefore, population specific normal values are needed for the quantitative determination of facial traits.

The Iranian Children with Down Syndrome

On the basis of the obtained frequencies, the facial traits in the collective of Iranian children with Down syndrome can be classified into three categories (Table 2):

1) ≥ 50% of the indices and angles outside p20 or p80: low midface (75%), narrow palpebral fissures (60%), laterally upslanting palpebral fissures (81%), short ears (62%) and anteriorly rotated ears (68%). These may be considered as characteristic facial traits of Iranian children with Down syndrome (n = 5).

2) ≥ 30% < 50% of the indices and angles outside p20 or p80: broad inner canthal distance (45%), prominent nose root (34%), short nose back (34%), everted nasal base (35%), long nasolabial distance (31%), forwards inclined integumental upper lip (35%), narrow mouth fissure (32 %), high chin (47%), prominent chin (38%), high-set ears (30%), narrow ears (47%) and narrow conchae (32%). These may be considered as additional facial traits of Iranian children with Down syndrome (n = 12).

3) < 30% of the indices and angles outside p20 or p80: interalar distance and maxillary prominence. These may be considered as unspecific facial traits of Iranian children with Down syndrome (n = 2).

Comparison with the Available Syndrome Knowledge

Three papers were identified in the literature, refering to anthropometric studies of the faces in Down syndrome:

In 1975 a small midface related to the endocranium in lateral cephalograms of a Caucasian Down syndrome population was defined (5). In our study it is reflected by a low midface height related to the bizygion diameter in 75% on frontal photographs of Iranian children with Down syndrome. They also identified a small mandible. This was different from the majority of literature reports, indicating a prognathic tendency in Down syndrome. In our study, a high chin related to the midface height (47%) and a prominent chin related to the ophryon radius (38%) were determined on profile fotographs of Iranian children with Down syndrome.

In 1985, facial measurements in 52 white North American individuals with Down syndrome was performed(4). Also midface hypoplasia was defined. The following facial features, comparable to our results, were found in 30% of the children or more: laterally upslanting palpebral fissures (46.9%; 81% in our results), short palpebral fissures (68,8; 60% in our results), short nose (40,4%; 34% in our results), short ears (65,4 and 71,5%; 62% in our results) and narrow ears (32,7 and 34,5%; 47% in our results). Trait definitions and frequency values, however, were different to our study.

In 1993, craniofacial measurements were performed on 199 (probably) Caucasian children and adults with Down syndrome and compared to age and sex-matched normal standards. An underdevelopment of the maxilla was found in comparison to the mandible, becoming increasingly apparent with age, as well as a reduced ear length. By a subset of three variables (ear length, maxillary arch and upper facial depth) they affected and unaffected individuals were distinguished (1).

Although these studies may contribute to an objective trait description in children with Down-syndrome, the results are difficult to compare because of the different methods and definitions used. The characteristic facial traits in Caucasian children with Down syndrome can not be considered to be definitively determined.

To compare the anthropometric results of the Iranian Down syndrome population a cohort of 42 unpublished German children with Down syndrome was used, which had been investigated with the same method as used in this study (Table 3). Concordances regarding the obtained frequencies were found for low midface (≥ 50%), short nose back, everted nasal base, long nasolabial distance, prominent chin and high set ears (≥ 30% < 50%). Three frequent “characteristic” features (≥ 50%) of the Iranian Down syndrome collective were among the less frequent “additional” features (≥ 30% < 50%) in the German cohort: narrow palpebral fissures, short ears, anteriorly rotated ears. Vice versa, two frequent “characteristic” features (≥ 50%) of the German Down syndrome cohort (≥ 50%) were among the less frequent “additional” features (≥ 30% < 50%) in the Iranian collective: broad inner canthal distance; narrow mouth fissure. Three less frequent “additional” features (≥ 30% < 50%) in the Iranian collective were among the rare, “unspecific” traits (< 30%) in the German cohort: prominent nose root; forwards inclined integumental upper lip; narrow ears. It is impossible to decide from the available data, whether these small differences are substantial and indicate different expressions of Down syndrome in both ethnically different populations, or whether they are due to other factors, e.g. the relatively small number of cases in the German syndrome cohort.

Objective metric definitions and frequency analyses of dysmorphic traits in children with genetic syndromes are necessary for scientifically founded syndrome descriptions in textbooks and databases. They form the basis of anthropological studies regarding individual phenotypic variabilities and their modifications at different ages and in different ethnic populations. Furthermore, it may be required for the construction of “phenotypic maps” regarding syndrome specific chromosomal regions by molecular marker analyses (7).

Table 1.**Variance (v) in ten photos of the same child, given as percentage of the Standard deviatiion (s) of the mean**

No.	Parameter	Mean	S	V
1	Midface height	24	1.05	4.3
2	Inner canthal distance	9	0.31	3.4
3	Width of palpebral fissures	6	o.31	5.1
4	Inclination of palpebral fissures	25	2.42	9.6
5	Depth of the nose root	9	0.79	8.7
6	Length of the nose back	22	1.05	4.7
7	Interalar distance	21	0.56	2.6
8	Inclination of the nasal base	42	0.52	1.2
9	Prominance of the upper jaw	17	0.97	5.7
10	Nasolabial distance	12	1.5	12.5
11	Inclination of the integumental upper lip	32	1.75	5.4
12	Width of the mouth	32	1.56	4.8
13	Height of the chin	16	1.26	7.8
14	Prominence of the chin	26	1.39	5.3
15	Inclination of the ear insertion line	29	2.48	8.5
16	Position of the ears	42	2.94	7
17	Length of the ears	23	0.69	3
18	Width of the ears	17	1.35	7.9
19	Width of the concha	49	2.54	5.1

Table 2.**Results of photoanthropometric measurements of 19 facial parameters in the collective of 136 Iranian children with Down syndrome in comparison with a collective of 100 normal Iranian children**

Paraneter		Age Effect	Down Syndrome			
		r	<p 20	%	>p 80	%
1	**Midface height**	0.33	Low	75	High	3
2	**Inner canthal distance**	-0.12	Narrow	2	Broad	45
3	**Palpebral fissure yidth**	0.17	Narrow	60	Broad	5
4	**Palpebral fissure inclination**	-0.06	Lat. downslanting	4	Lat. upslanting	81
5	**Depth of the nose root**	-0.54	Prominent	34	Sunken	16
6	**Length of the nose back**	0.60	Short	34	Long	29
7	**Inclination of the nasal base**	0.45	Inverted	8	Everted	35
8	**Interalar distance**	0.23	Narrow	7	Broad	27
9	**Maxillaiy prominence**	0.42	Prominent	26	Receding	24
10	**Nasolabial distance**	-0.30	Short	10	Long	31
11	**Integ. upper lip inclination**	0.001	Backwards	7	Forwards	35
12	**Mouth width**	0.26	Narrow	32	Broad	11
13	**Chin height**	0.01	Low	15	High	47
14	**Chin prominence**	0.48	Receding	21	Proninent	38
15	**Ear position**	-0.42	High-set	30	Low- set	26
16	**Ear inclination**	0.12	Anteriorly	68	Posteriorly	3
17	**Ear length**	-0.23	Short	62	Long	4
18	**Ear width**	-0.27	Narrow	47	Broad	10
19	**Concha width**	-0.23	narrow	32	broad	13

Table 3. **Comparison of the anthropometric results from the Iranian collective (n = 136) and those from a German cohort of children with Down-syndrome (n = 42)**

Down syndrome	**Iranian**	**German**
concorances (≥ 50%)	(%)	(%)
low mid face	75	54
Concordances (≥ 30% < 50%)	**%**	**%**
short nose back	34	39
everted nasal base	35	42
long nasolabial distance	31	39
prominent chin	38	48
high set ears	30	46
discordances (≥ 50%)	**%**	**%**
narrow palpebral fissures	60	31
short ears	62	43
anteriorly rotated ears	68	30
broad inner canthal distance	45	66
narrow mouth fissure	32	57
discordances (≥ 30% < 50%)	**%**	**%**
prominent nose root	34	28
forwards inclined integumental upper lip	35	28
narow ears	47	26

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