Coll. Antropol. **27** Suppl. 2 (2003) 23–30
UDC 572.545:616-056.7
Original scientific paper

Craniofacial Anthropometric Analysis in Down's Syndrome Patients

Ivana Bagić and Željko Verzak

Department of Pedodontics, School of Dental Medicine, University of Zagreb, Zagreb, Croatia

A B S T R A C T

Past investigations of Down's syndrome (DS) have indicated that there are marked abnormalities in the craniofacial morphology. The aim of this study was to establish the craniofacial anthropometric variables which discriminate DS group from healthy population and also to observe the changes occuring with growth. Using noninvasive method of craniofacial anthropometry, the craniofacial pattern profile (CFPP) analysis (from twenty-five anthropometric measurements per person) was performed in 104 DS individuals and 365 healthy controls, aged seven to fifty-seven and divided into four age ranges. Z-scores were calculated for each variable and the variations in the craniofacial region have been identified by multivariate discriminative analysis. The results showed that three variables (head length (g-op), head circumference (OFC) and outer canthal distance (ex-ex)) were responsible for 85.68% variability ($p<0.001$). The analysis of z-scores showed that the majority of variables were in subnormal (under –2 SD) and normal range (from –2SD to +2SD), but none of them was in the supernormal range (over the +2SD). Some craniofacial characteristics are age-related. On the basis of craniofacial anthropometric traits it was posssible to separate even 91.35% of DS patients from the healthy population. It could be concluded that these findings demonstrate the usefulness of application of CFPP in defining abnormal craniofacial dimensions in DS individuals.

Key words: *Down syndrome, pattern profile analysis, craniofacial measurements*

Introduction

More than a hundred signs have been described in the literature about Down syndrome[1,2]. Disagreement in syndrome identification is common, and diagnostic accuracy would be enhanced by objective quantitative criteria and analytic meth-

Received for publication April 28, 2003

odology where possible. Craniofacial anthropometry provides a simple and non-invasive method of quantitative assessment of changes in the surface anatomy of the head and the face in individuals with Down syndrome. Anthropometry can be greatly enhanced by the production of pattern profiles after converting individual measurements into z-scores, because any dimension characterised by a low or high z-score is immediately obvious, it is of potential diagnostic value and serves to identify most deviant values of craniofacial parts[3–5]. Previous studies of Down syndrome showed signs of facial differences based only on visual examination of the head and the face, or focused on only one region of the craniofacial complex[6,7]. Some other studies evaluated the morphological changes covering only some aspects of craniofacial defects[8,9]. More recent Canadian studies investigated age-related changes of not only linear and angular measurements but the indices in the craniofacial regions. Craniofacial indices are usually used to illustrate the relationship between individual measurements and thus the main proportion qualities of the face and head[10,11]. In Croatian Down syndrome population there were no data about craniofacaial anthropometric measurements.

The purpose of this study was to identify the normal and abnormal (subnormal or supernormal) craniofacial anthropometric measurements which contribute to the craniofacial stigmata of individuals with Down syndrome and to demonstrate the age-related changes in the findings.

Patients and Methods

One hundred four Caucasian individuals with Down syndrome, age 7 do 57 years and divided into four age ranges (7 to 12, 13 to 18, 19 to 29 and 30 to 57) were chosen as a defined syndrome population to investigate the development and application of anthropometric craniofacial pattern profiles. Down syndrome patients were drawn from Primary school for children with developmental disturbances »Nad lipom«, Centers for handicapped or abandoned children »Sloboština«, »Paunovac«, »Ilica« and Center for handicapped persons »Stančić«. The control group consisted of 365 age- and sex-matched healthy individuals.

A total of twenty-five craniofacial measurements per patient were performed by one measurer, following the methodology according to Farkas, using an non-elastic tape, and spreading and sliding callipers[12]. In a cooperative patient, the complete measurement took 20 to 30 minutes. Specific craniofacial variables were chosen to represent dimensions of craniofacial widths, lengths, depths, heights, and circumference (Table 1).

The data were compared to dimensions of 365 age- and sex-matched healthy individuals and converted to z-scores to control the age and sex differences. Pattern profiles were compiled for each age and sex, and discriminant function analysis was used to separate Down syndrome subjects from normal individuals (Statistical Package for Social Scientists (SPSS)).

Results

Abnormal measurements were qualified as subnormal if they were smaller than the mean value minus 2 standard deviations (–2SD) of the controls. Likewise, supernormal measurements exceeded the value of the mean plus 2SD (+2SD) of the controls, while normal measurements were between minus 2SD to plus 2SD (from –2SD to +2SD). In all Down syndrome measurements there was no variable which was in supernormal range compared to subnormal range. The most common subnormal measurements were taken as follows: head length

TABLE 1
ANTHROPOMETRIC MEASUREMENTS EMPLOYED IN THIS STUDY

Head width	eu-eu	Eurion to eurion
Skull base width	t-t	Tragion to tragion
Minimum frontal width	ft-ft	Frontotemporale to frontotemporale
Upper facial width	zy-zy	Zygion to zygion
Lower facial width	go-go	Gonion to gonion
Head length	g-op	Glabella to opisthocranion
Upper facial depth-dex	n-t-dex	Right tragion to nasion
Midfacial depth-dex	sn-t-dex	Right tragion to subnasion
Lower facial depth-dex	gn-t-dex	Right tragion to gnation
Upper facial depth-sin	n-t-sin	Left tragion to nasion
Midfacial depth-sin	sn-t-sin	Left tragion to subnasion
Lower facial depth-sin	gn-t-sin	Left tragion to gnation
Total facial height	n-gn	Nasion to gnation
Upper facial height	n-sn	Nasion to subnasion
Nasal width	al-al	Alare to alare
Mouth width	ch-ch	Cheilion to cheilion
Inner canthal distance	en-en	Endocanthion to endocanthion
Outer canthal distance	ex-ex	Exocanthion to exocanthion
Ear width-dex	pra-pa-d	Preaurale to postaurale right
Ear length-dex	sa-sba-d	Superaurale to subaurale right
Ear width-sin	pra-pa-s	Preaurale to postaurale left
Ear length-sin	sa-sba-s	Superaurale to subaurale left
Maxillary arc	t-sn-t	Tragion to subnasion to tragion
Mandibular arc	t-gn-t	Tragion to gnation to tragion
Head circumference	OFC	Maximum circumference in horizontal plane at level of glabella and opisthocranion

(g-op), outer canthal distance (ex-ex), ear width (pra-pa dex and sin), ear length (sba-sa dex and sin) and head circumference (OFC) in 57.7 to 86.5% DS patients. Next six variables were in subnormal values presented in 32.7–46.2% DS patients: upper facial depth (n-t), midfacial depth (sn-t), upper facial height (n-sn) and maxillary arc (t-sn-t).

Figure 1 compares the pattern profiles in males (fine line) and females (dotted line) with DS. All profiles display z-scores on the vertical axis and craniofacial dimensions on the horizontal axis. The profiles represent a fairly similar appearance. Almost all dimensions were slightly higher in males than in females, except in three dimensions (g-op, pra-pa-dex and pra-pa-s). All dimensions were in normal (–2SD to +2SD) or subnormal range (under –2SD) which means that these variables which are in subnormal range (g-op, n-t-dex, sn-t-dex, n-sn, ex-ex, pra-pa-d, sa-sba-d, sa-sba-s and OFC) are abnormally small compared to the control group. None of the dimensions was in the supernormal range. The comparison of all male age groups showed that there were no dimensions in supernormal range (over +2SD), and all variables were in normal (–2SD to +2SD) and subnormal (under –2SD) range (Figure 2). All male age groups had the same four variables in the subnormal range (under –2SD): head length (g-op), ear width (pra-pa), ear

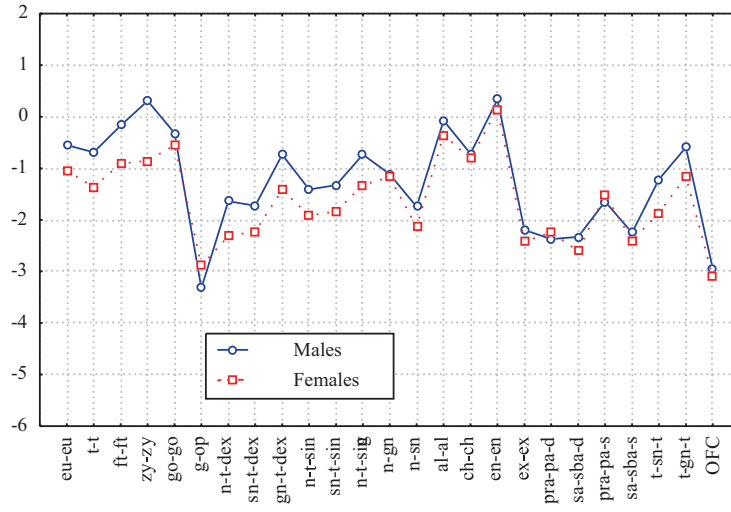

Fig. 1. A craniofacial pattern profile comparing males and females with Down syndrome.

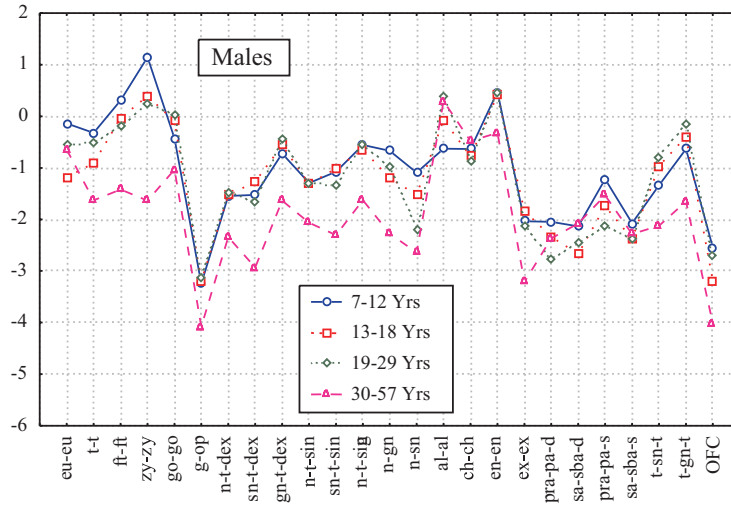

Fig. 2. A craniofacial pattern profile comparing all four age groups (7 to 12, 13 to 18, 19 to 29 and 30 to 57 yrs) in Down syndrome males.

length (sa-sba) and head circumference (OFC). The first, youngest group (7 to 12 yrs) had five dimensions in the subnormal range (under –2). The second age group (13 to 18 yrs) also had five dimensions in the subnormal range (under –2). The third age group (19–29 yrs) had eight dimensions in the subnormal range (under –2). The oldest group (30 to 57 yrs) also had eight dimensions in the subnormal range (under –2). They were: head length (g-op), upper facial depth (n-t), midfacial depth (sn-t), total facial height (n-gn), upper facial height (n-sn), outer

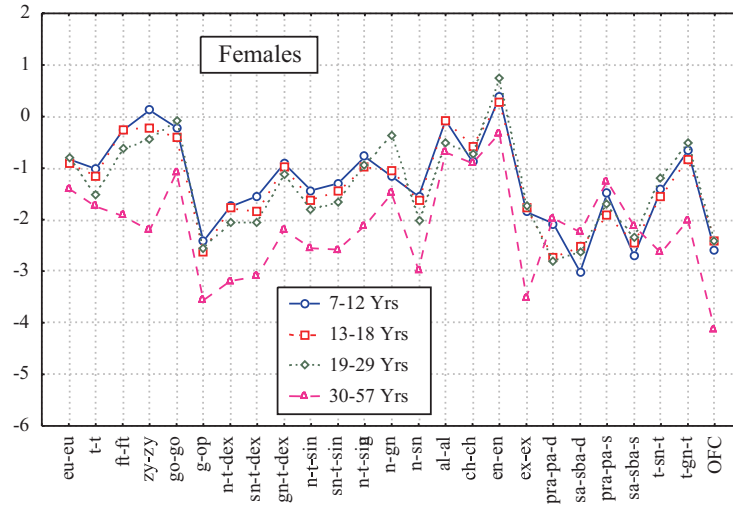

Fig. 3. A craniofacial pattern profile comparing all four age groups (7 to 12, 13 to 18, 19 to 29 and 30 to 57 yrs) in Down syndrome females.

canthal distance (ex-ex), ear length (sa-sba), maxillary arc (t-sn-t) and head circumference (OFC).

The comparison of all female age groups showed that there were no dimensions in the supernormal range (over +2SD), and all variables were in the normal (–2SD to +2SD) and subnormal range (under –2SD) (Figure 3). All female age groups had head length (g-op), ear width (pra-pa), ear length (sa-sba) and head circumference (OFC) in subnormal range (under –2), the same as in male groups. The youngest group (7 to 12 yrs) had five dimensions in the subnormal range (under –2). The second age group (13 to 18 yrs) had five dimensions in the subnormal range (under –2). The third age group (19–29 yrs) had eleven dimensions in the subnormal range (under –2). The oldest group (30 to 57 yrs) also had eleven dimensions in the subnormal range (under –2). They were: upper facial width (zy-zy), head length (g-op), upper facial depth (n-t), midfacial depth (sn-t), lower facial depth (t-gn), upper facial height (n-sn), outer canthal distance (ex-ex), ear length (sa-sba), maxillary arc (t-sn-t), mandibular arc (t-gn-t) and head circumference (OFC).

From the total number of Down syndrome subjects, a discriminant analysis showed that even 91.35% individuals were accurately classified which means that there are obvious characteristics which separate them from the healthy population.

Discussion and Conclusions

The identification of severely abnormal findings enables us to show the most striking deviations from the normal ones. This study focused attention on the signs of the face and head that are most frequently described in the Down syndrome literature[1]. Our data confirmed the traditional knowledge of head and facial appearance in Down syndrome population by means of the most frequent subnormal measurements. These are: head length (g-op), outer canthal distance (ex-ex), ear width (pra-pa dex and sin), ear length (sba-sa dex and sin) and head circumfer-

ence (OFC) (frequencies of 57.7–86.5%). So, these findings are confirming the most frequently described brachycephaly, round face and small ears in Down syndrome population[13]. The next six variables also showed subnormal values, but presented in lower frequencies (32.7–46.2%) of Down syndrome patients, confirming an underdevelopment of maxilla in comparison with mandibula. These six subnormal values were: upper facial depth (n-t), midfacial depth (sn-t), upper facial height (n-sn) and maxillary arc (t-sn-t). In all Down syndrome measurements there was no variable in supernormal range, but only in normal and subnormal range. In Farkas's study, supernormal findings were encountered, but far less frequently than subnormal, and were not found at all in the ears[14]. Severe degrees of supernormality were manifested most often in obtuse mentocervical angles, increased tilts of palpebral fissure line which variables weren't measured in this study[14]. The comparison of all male and female age groups showed that there were no dimensions in supernormal range (over +2), and all variables were in normal (–2 to +2) and subnormal (under –2) range. The lack of supernormality in our study could be influenced by the size of the study group as well as by variations in ethnic differences between ours and other studies.

Some observations in our study confirmed many of those in the literature but many did not[15–21]. Previous publications on craniofacial morphology in Down syndrome patients of Croatian origin did not exist. Also, this investigation could not be completely compared with others because of the ethnic origin, age ranges or various statistical interpretations. Farkas and al. showed in their study an age range distribution different from ours. Their oldest Down syndrome patient was 36 years old, and ours was 57. He divided his patients into three study groups and found that the growth rate in the youngest Down syndrome patients (1 to 5 yrs) was below the level of the healthy population, approached it in age group 6 to 15 (coinciding with the period of maturation), and rose slightly in some measurements in age group 16 to 36[14]. Our investigation showed that in males and in females there are first two aged groups (7 to 12 and 13 to 18 yrs) where a lower number of variables in subnormal range are presented, compared to the older ones (19 to 29 and 30 to 57). Beside that our data showed that the number of variables in subnormal range is increasing greater with age.

There are many different interpretations of frequency of abnormal findings in the craniofacial complex and ours is just one more proof of some variabilities in interpretations of accurate stigmata of Down syndrome individuals. Obviously, there is a need for a larger, more representative sample to resolve some differences in a variety of ethnic and age structure classification interpretations encountered in the literature.

This study showed the use of pattern profiles and their application to Down syndrome diagnosis. Pattern profiles provide a technologically simple adjunct to our subjective evaluation. Z-score conversion and pattern profile generation is computerised and the normal and unusual (subnormal and supernormal) dimension are easily visualised. Subjective impressions can be validated by statistical techniques and discriminant function analysis can complete the diferentiation of Down syndrome subjects.

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I. Bagić

Department of Pedodontics, School of Dental Medicine, University of Zagreb, Gundulićeva 5, 10000 Zagreb, Croatia

KRANIOFACIJALNA ANTROPOMETRIJSKA ANALIZA U PACIJENATA S DOWNOVIM SINDROMOM

SAŽETAK

Ranija istraživanja pokazala su kako postoje naglašene abnormalnosti u kraniofacijalnoj morfologiji Downovog sindroma (DS). Cilj ovog istraživanja bio je utvrditi kraniofacijalne antropometrijske varijable koje diskriminiraju osobe s DS u odnosu na zdravu populaciji i promotriti promjene tijekom rasta. Pomoću potpuno neinvazivne metode kraniofacijalne antropometrije napravljena je analiza kraniofacijalnog antropometrijskog profila (KAP) (na temelju dvadeset antropometrijskim varijabli izmjerenih na svakoj osobi) i to kod 104 osobe s DS i 365 zdravih osoba, u dobi od sedam do pedeset sedam godina, podijeljenih u četiri dobna razreda. Standardizirane vrijednosti (z-vrijednosti) izračunate su za svaku varijablu, a varijacije u kraniofacijalnoj regiji utvrđene multivariatnom diskriminativnom analizom. Rezultati su pokazali da su tri varijable (dužina glave g-op, opseg glave opseg i vanjska kantalna udaljenost ex-ex) odgovorne za 85.68% varijabiliteta ($p<0.001$). Analiza Z-vrijednosti pokazala je da je većina varijabli u subnormalnom (ispod –2SD) i normalnom (od –2SD do +2SD), a niti jedna u supernormalnom (iznad +2SD) području. Neke kraniofacijalne karakteristike ovisne su o dobi. Na temelju kraniofacijalnih antropometrijskih obilježja moguće je toč-

no izdvojiti čak 91.35% osoba s DS u odnosu na zdrave osobe. Moglo bi se zaključiti da ovi nalazi pokazuju korisnu primjenu KAP-a u definiranju abnormalnih kraniofacijalnih dimenzija u osoba s DS.

Ključne riječi: Downov sindrom, kraniofacijalni antropometrijski profil, kraniofacijalna njerenja