A genetic study of anteroposterior and vertical facial proportions using model-fitting

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In the literature, different heritabilities have been reported for facial characteristics and dentofacial variables. Variations between 20% and 90% are found. On average, higher heritabilities have been found for vertical variables than for horizontal and dental ones.[1,2] The genes that determine facial characteristics appear to act in an additive manner, except for mandibular length, which is inherited by dominant alleles.

It has been suggested that the relative impact of genes and environment on facial composition differs from the genetic control of facial components.[3] That is why some investigators found low heritabilities for the whole craniofacial complex, but high heritabilities for contours of the individual bones.[4,5]

Therefore, the aim of this study was to evaluate the genetic and environmental contribution to facial proportions and to compare them with earlier genetic analyses of the different facial components using model-fitting and path analysis.

Materials and methods

Seventy-nine pairs of twins between 9 and 16 years old volunteered to participate in this study. All were from the East Flanders Prospective Twin Survey, a survey that includes all multiple births in East Flanders since 1964.[6]

Monochorial twins were considered as monozygotic (17 twin pairs); dichorial twins of unlike sex (18 twin pairs) were considered as certainly dizygotic. Of the remaining same-sex dichorial twins, bloodgroups and five DNA polymorphisms were determined. If at least one genetic difference was found, the twins were considered dizygotic (28 twin pairs). Those with identical genetic markers were considered

Abstract

Genetic model-fitting was used to determine the heritability of anteroposterior and vertical facial proportions in twins. Lateral headplates of 33 monozygotic and 46 dizygotic twins, none of whom had undergone orthodontic treatment, were used. Five proportions, based on four vertical and five horizontal measurements, were assessed: lower facial height, anterior- to posterior-facial height, total facial height to face depth, sella–A-point to sella–B-point, and sella–upper incisal edge to sella–lower incisal edge. Reproducibility was high for all variables. Model-fitting indicated that all the facial proportions were controlled by additive genes and the specific environment. The genetic component was 71% for upper- to lower-facial height, 66% for anterior- to posterior-facial height, 62% for total facial height, and 66% for sella-A-point to sella-B-point and sella-upper incisal edge to sella-lower incisal edge.

Key words

Facial proportion • Genetics • Twins

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Figure 1
Cephalometric distances used for studying the heritability of facial proportions
Vertical proportions (parallel to X-axis):
ANSV / ANSGNV
GNV / GOV
Horizontal proportions (parallel to Y-axis):
ISH / IIH
AH / BH
Vertical/horizontal proportion:
GNV / POGH

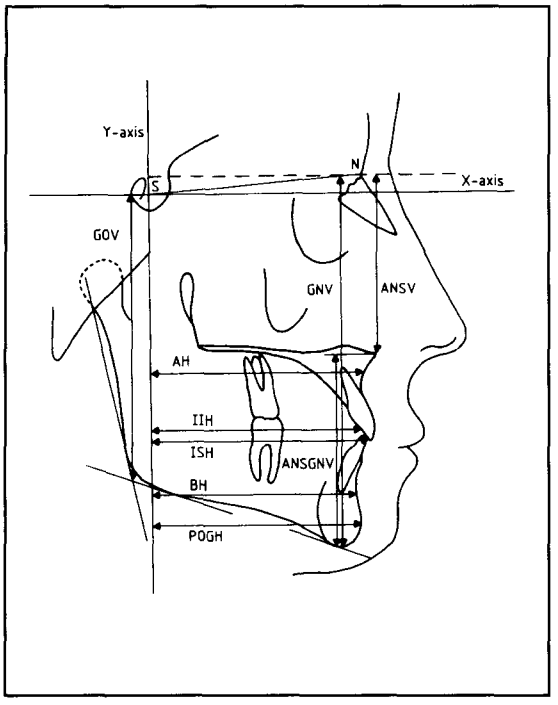

Figure 1

monozygotic (16 MZ twin pairs); their probability was calculated[7] and found to be at least 99%.

Standard lateral headplates were taken of the 33 MZ (17 male and 16 female) and the 46 DZ twin pairs (14 male, 14 female and 18 male-female). The location of the landmarks and definitions of the landmarks and reference lines are according to Rakosi[8] and Lundström and McWilliam.[1] Reference points were digitized[9,10] and nine distances were calculated automatically with the Quick Ceph program (Orthodontic Processing Co, Chula Vista, Calif) on a Mac II computer.

The variables studied are listed in Table 1 and presented in Figure 1. They consist of five proportions based on four vertical and five horizontal measurements used in another study.[2] Mean, standard deviation, and range are shown in Table 1.

Measurement error

To determine the measurement error, measurements were performed twice on the lateral headplates by the same person, with one month interval, on 10 randomly selected twin pairs. The means and variances of the first and second measurement and of the differences between them were compared.[11] The error variance Se^2 was calculated with the Dahlberg[12] formula ($Se^2=\Sigma d^2/2n$), where d is the difference between the two repeated measurements.

Table 1
Age distribution, mean, standard deviations, and range of the variables

Variable	Mean	S.D.	Range
Age	12.1 y	/	9-16 y
ANSV/ANSVGNV	0.81	0.06	0.77-0.85
GNV/GOV	1.65	0.10	1.56-1.71
ISH/IIH	1.06	0.03	1.04-1.08
AH/BH	1.21	0.09	1.14-1.28
GNV/POGH	2.29	0.37	2.06-2.53

Statistical analysis

The statistical analysis was done with SAS (Statistical Analysis System, version 6.08). Probabilities below 0.05 were considered statistically significant.

Genetic analysis by model-fitting was done with MX.[13,14] Path analysis allows one to estimate the significance of different components of variance, i.e., the additive (A) and dominant (D) genetic factors and the common (C) and unique (E) environmental factors. Different models, each representing a different hypothesis, were tested in a univariate way.

The AE model supposes that the variance consists of only two factors, i.e., an additive genetic and a unique environmental component. The models that include dominance or common environment are represented respectively as ADE and ACE. The CE model hypothesizes that there is no genetic influence at all, but only an environmental one, common to both members of each pair; while the E model supposes that the variance is solely caused by the unique environment, specific to each member of the pair. The power of the sample was sufficient to detect additive genetic influence, but might be too low to detect dominance or shared environment, unless the effect was large.

To test whether the genetic and environmental effects were the same in different sexes, gender heterogeneity was tested by estimating different parameters for genetic and environmental factors in males and females (gAE or gACE).

Goodness of fit was evaluated by χ^2. This statistically represented the significance of the difference between the observed data and the data expected on the basis of each hypothesis: The lower the χ^2, the better the fit. When the prob-

Table 2
Heritability and environmental estimates

Variable	Model-fitting							Heritability according to Rao et al.(1976)							
	Genetic Environment additive specific			Genetic Environment additive common specific				This study Correlation				Lundström 1988 Correlation			
	Model	a^2	e^2	Model	a^2	c^2	e^2	MZ	DZ	h^2	c^2	MZ	DZ	h^2	c^2
Vertical proportion															
ANSV/ANSGNV	AE	0.71	0.29	ACE	0.60	0.01	0.29	0.75	0.34	0.81	-0.06	0.77	0.48	0.58	0.19
GNV/GOV	AE	0.66	0.34	ACE	0.34	0.29	0.37	0.65	0.43	0.44	0.21	0.68	0.26	0.84	-0.16
Horizontal proportion															
ISH/IIH	AE	0.62	0.38	ACE	0.30	0.29	0.41	0.64	0.52	0.25	0.40	0.49	0.16	0.66	-0.17
AH/BH	AE	0.66	0.34	ACE	0.34	0.30	0.36	0.61	0.43	0.36	0.25	0.87	0.38	0.82	-0.11
Vertical:horizontal proportion															
GNV/POGH	AE	0.66	0.34	ACE	0.30	0.33	0.37	0.66	0.34	0.64	0.02	0.82	0.41	0.82	0.0

$h^2 = 2(r_{imz} - r_{idz})$; $c^2 = 2r_{idz} - r_{imz}$

ability (p) of χ^2 was > 0.05, the fit of the model was acceptable. The Akaike information criterion[15] (AIC), which combined χ^2 with degrees of freedom (AIC=χ^2 -2df), gave an idea of the parsimony of a model. For each variable, the model with the lowest AIC was considered the best-fitting model. All models were compared with the model involving only the specific environment. The difference between their goodness of fit χ^2 was used to test whether the new model was significantly better than the E model. The sum of the squared estimates for each model was equal to 1.

In order to compare our data with the data of Lündström and McWilliam,[3] heritability values were calculated according to Rao et al.[16]

Results

Measurement error

The measurement error was small. No significant differences were found between the means of the first and the second measurements. Reproducibility was high.[2]

Heritability estimates

For all facial proportions, the best-fitting model was the AE model, although the ACE model was not significantly worse (p>0.05). Heritabilities ranged between 62% (ISH/IIH) and 71% (ANSV/ANSGNV), with somewhat higher heritabilities for the horizontal proportions than for the vertical ones (Table 2).

The proportion of common environmental influence estimated from the less-fitting ACE models amounted to approximately 30%.

Discussion

In order to compare the results with those of Lundström and McWilliam,[3] variables were divided into vertical, horizontal, and vertical versus horizontal proportions. The AIC values for each model were calculated and the results of the most parsimonious best-fitting model were retained. All retained models fitted well. The estimated parameters were not different between the sexes.

For all facial proportions, the best-fitting model was the AE model, although the ACE model was not significantly worse. Heritabilities ranged between 62% (ISH/IIH) and 71% (ANSV/ANSGNV), with somewhat higher heritabilities for the horizontal proportions than for vertical ones. As mentioned above, our sample size might have been too small to prove that environmental factors common to both twins—such as diet or the use of a pacifier—had a significant influence. The proportion of common environmental influence estimated from the less-fitting ACE models amounted to approximately 30%. The inclusion of common environment decreased the amount explained by genetic factors.

Because Rao's[16] technique and model-fitting with MX to calculate heritabilities are fundamentally different, a comparison is difficult. When the heritability estimated by the two different methods on the same data are compared, it is striking that the genetic determination based on Rao's[14] formula is mostly lower than that calculated by model-fitting, except for the first vertical proportion. Except for the ANSV/ANSGNV, the heritabilities as estimated by Lundström and McWilliam[3] are higher than or similar to those calculated by the same formula on our data.

For ISH/IIH the results are similar to those of Lundström and McWilliam,[3] who found the lowest genetic dependence for this anteroposterior incisal edge relationship.

Conclusions

High genetic determination was found for the vertical proportions. The lowest heritability values were found for sella-upper incisal edge to sella-lower incisal edge.

The lower genetic determination of the facial proportions compared with their components indicates that the indices are subject to more complex interactions between genes and environment.

Variables with a lower genetic determination are more open to influence by, for example, orthopedic correction than are variables with a high genetic determination, which are not so easily changed by the environment.

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