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Genetic Contribution to Variation in Body Configuration in Belgian Nuclear Families: A Closer Look at Body Lengths and Circumferences

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A B S T R A C T

The purpose of the current study was to evaluate the contribution of genetic factors on body configuration related phenotypes. The sample consisted of 119 Belgian nuclear families including 231 males and 229 females. Factor analysis with varimax rotation was carried out to analyse 13 length and circumference measures and the resulting two synthetic traits (LF and CF; linear and circumference factors, respectively) were used as summary variables. Univariate quantitative genetic analysis indicated that variation in anthropometric as well as in synthetic traits was significantly dependent on additive genetic effects, with heritabilities ranging from 0.55 to 0.88. Narrow sense heritability estimates were higher for measurements principally characterizing skeletal mass than in variables that also involve soft-tissues. Sex, age and their interactions explained 11–67% of the total phenotypic variance. This report also examined the covariations between pairs of anthropometric and synthetic traits (length measurements and LF vs. height; circumference measures and CF vs. *weight and BMI; LF* vs. *CF). Significant genetic correlations among all the studied traits (except for middle finger length* vs. *height) confirmed the influence of pleiotropy on genetic determination of these phenotypes. Bivariate analysis showed that pleiotropic effects had a great influence in determining body traits variation within body length measurements, as well as between body circumferences and weight or BMI. In relation to the two synthetic traits, even the variation of body lengths and circumferences was highly determined by genetic factors, shared genetic influences were unlikely to explain much of the observed variation between LF and CF. The results of the present study allow us to conclude that in this population body configuration related traits are subject to a strong genetic control and that shared genes also contribute to this genetic structure.*

Key words: *variance components, heritability, anthropometry, factor analysis, genetic correlations*

Introduction

Anthropometric phenotypes are determined by multiple genes, environmental factors and interactions among them, leading to a continuous distribution of phenotypic values. Genetic factors exert their influence by an unknown number of genes each of which depends on an unknown number of alleles[1]. On the other hand, environmental factors are very variable and include sources of variation as nutrition, intrauterine environment, family size and socioeconomic-status[2]. The existence of considerable genetic influence on the variation of anthropometric traits is widely accepted, and is supported by several twin[3–6], adoption[7,8] and family studies[9–12]. Although these genetic and environmental influences have been extensively studied, the strength of their contributions to these phenotypes is not completely defined.

Until now, the great majority of these inheritance studies have been carried out on height, weight and body mass index (BMI)[6,11,13–17], obtaining heritability estimates higher than 60% for these three phenotypes. Relatively few investigations have focused on the heritability of other body configuration traits and even less frequents are the studies employing principal component or factor analysis[18–22]. Although the study of genetic determina-

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tion of individual morphological traits is of substantial interest, factors extracted from a factor analysis could represent features that contain a higher degree of genetic variance than the original variables separately[23]. Furthermore, factor analysis reduces a large set of measures into a few uncorrelated factors and can be employed as a complementary approach that may provide a more general view of human body configuration.

Despite the significant influence of genetic effects on the determination of anthropometric traits and the assumed existence of shared polygenic effects (i.e. pleiotropy) between body morphology determinants, to our knowledge, hardly any studies have focused on pleiotropic effects between anthropological body measures. Livshits et al.[21] in an attempt to discover the genetic determinants responsible for the variability and covariability of the adiposity traits conclude that pleiotropic gene effects play an important role in their variation. In addition, Choh et al.[24] in a study conducted in a population of adult Samoans, found significant genetic and environmental correlations among measures of body fatness. Thus, further research is needed in this field to lay down the basis of the genetic and environmental determinants responsible for the overall body configuration.

In the present study, a variance component analysis was conducted to explore the genetic structure of several anthropometric phenotypes in Belgian population using nuclear family data. The major aims were (1) to estimate the contribution of the genetic effects (h^2) on body configuration related traits (2) to evaluate the confounding effects of possible covariates on these phenotypes and (3) to explore the degree to which the studied pairs of traits were influenced by the same genetic and environmental factors.

Materials and Methods

Sample

The sample consisted of 460 individuals (231 males and 229 females) from 119 nuclear families comprising a marital couple and at least one child. Age range varied from 17 to 72 years (mean age 38.06) in males and from 17 to 68 years (mean age 36.58) in females. The sample was taken between the years 1965–1967 and is not representative of the whole Belgian population. It is both geographically selected, because the subjects were Belgians living in Brussels, and also socially selected, the children were mainly students of high schools of Brussels. The data were collected after receiving consent from the members of the families included in the study. Analyses with these data have been published in various works[9,25], and have been reanalysed for the current study with a different methodology (adjustment for more covariates and variance decomposition method) and new approaches (factor analysis and bivariate analysis).

Measurements

Using the standard methods of measurement[26] the following anthropometric traits were taken by the same investigator (C.S.) from each participant: height (cm), weight (kg), sternal height (cm), sitting height (cm), arm length (cm), upper arm length (cm), forearm length (cm), middle finger length (cm), neck circumference (cm), upper arm circumference (relaxed) (cm), upper arm circumference (contracted) (cm), wrist circumference (cm), calf circumference (cm) and ankle circumference (cm). In addition, BMI was calculated from height and weight measures [weight (kg)/height (m^2)]. All length variables were measured using a Siber-Hegner anthropometer (GPM, Switzerland) accurate to one millimetre. Body circumferences were measured to the nearest 1mm with a measuring tape and body weight was measured using a beam balance (accuracy of 0.1 kg).

Preliminary statistical analysis

The basic descriptive statistics (mean, standard deviation, maximum and minimum) were carried out for age and anthropometric traits separated by sex. Next, length and circumference traits were subjected to a factor analysis with orthogonal varimax rotation. The eigenvalue of 1 criterion was implemented to retain the factors, and scores for each individual on the extracted two factors (Linear Factor, LF and Circumference Factor, CF) were used in further analysis. These preliminary statistical computations were conducted using the SPSS package version 14.0 for Windows (SPSS Inc., Chicago, IL, USA).

Quantitative genetic analysis

Under a pure polygenic model, the phenotype (P) is a function of genetic (G) and environmental (E) effects (i.e., P=G+E), usually expressed in terms of variance components ($V_P=V_G+V_E$). Genetic component is further divided into genetic additive, dominance, and interaction variances whereas the environmental component includes factors such as socioeconomic status and acculturation. Narrow sense heritability (h^2) is defined as the proportion of the total phenotypic variance of a trait attributable to additive genetic sources of variation.

In order to evaluate the contribution of these additive genetic effects on inter-individual variation of the analysed traits, variance component analysis was performed using the SOLAR program (Sequential Oligogenic Linkage Analysis Routines), available online (http:/www.sfbr.org/sfbr/public/software/solar/solar.html). This analysis partitions the observed phenotypic variance into additive genetic and environmental components, by maximum-likelihood methods[28]. Significance of heritability estimates was tested formally by comparing the likelihoods for the restricted model, in which each of the parameters was constrained to zero, to the likelihood for the general model, in which all parameters were estimated. The likelihood ratio test is asymptotically distributed as a χ^2, with the degrees of freedom being given by the difference in the number of parameters estimated in the two

models[27]. Simultaneously with the narrow-sense heritability, the effects of age, sex and their interactions were estimated in these analyses in order to increase the accuracy of the heritability estimates by decreasing the proportion of the residual phenotypic variation attributable to random environmental factors.

Finally, using the information obtained from univariate analyses, bivariate variance components analysis was undertaken to determine the extent to which shared genetic and environmental effects influence the covariation among pairs of anthropometric and synthetic traits. Again, the SOLAR program was used in these procedures. This method decomposes the phenotypic correlations (ρ_P) between pairs of traits into the underlying genetic (ρ_G) and environmental correlation (ρ_E), correcting for the use of related individuals, by the following equation:

$$\rho_P = [\rho_G \cdot \sqrt{h_1^2} \cdot \sqrt{h_2^2}] + [\rho_E \cdot \sqrt{(1-h_1^2)} \cdot \sqrt{(1-h_2^2)}]$$

where h_1^2 and h_2^2 are respective heritabilities of trait 1 and trait 2[29]. A more detailed explanation of this methodology can be found in a previously published work[30]. Significance of both ρ_G and ρ_E between any pair of traits was tested by the likelihood ratio test and complete pleiotropy was assessed by comparing the general model, against a restricted model in which ρ_G was constrained to 1.0. In addition, in order to assess the contribution of pleiotropic genetic effects on the variation of these traits, bivariate heritability was estimated as ρ_G^2, as shown in Yang et al.[31].

TABLE 1
DESCRIPTIVE STATISTICS FOR STUDIED TRAITS IN BELGIAN SAMPLE

Traits		Valid *n*	Min.	Max.	$\overline{X}$	SD
Age (years)	Males	231	17.0	72.1	38.06	16.44
	Females	229	17.2	68.1	36.58	14.88
Height (cm)	Males	230	155.6	194.4	174.34	6.62
	Females	229	146.0	181.6	162.38	5.84
Sternal height (cm)	Males	230	125.1	159.3	142.01	5.86
	Females	227	118.1	149.7	132.02	5.36
Sitting height (cm)	Males	230	82.2	99.3	90.65	3.46
	Females	228	78.8	96.4	85.65	2.92
Arm length (cm)	Males	230	69.0	88.4	77.37	3.75
	Females	229	63.7	80.6	70.94	3.39
Upper arm length (cm)	Males	231	27.7	38.4	32.81	2.07
	Females	229	25.0	35.2	30.25	2.00
Forearm length (cm)	Males	230	22.0	29.9	25.62	1.42
	Females	228	19.4	26.9	23.21	1.28
Middle finger length (cm)	Males	228	9.0	11.5	10.07	0.51
	Females	228	8.0	10.9	9.22	0.52
Neck circumference (cm)	Males	230	31.5	43.8	36.88	2.08
	Females	228	28.5	37.5	32.19	1.79
Upper arm circ. (rel.) (cm)	Males	231	22.2	36.2	28.58	2.70
	Females	229	19.9	38.4	27.64	3.06
Upper arm circ. (cont.) (cm)	Males	231	22.4	37.0	29.45	2.81
	Females	229	20.2	38.5	27.96	3.09
Wrist circumference (cm)	Males	228	14.7	20.1	17.03	0.83
	Females	229	13.3	19.0	15.26	0.87
Calf circumference (cm)	Males	229	28.7	41.3	35.58	2.30
	Females	228	27.9	41.0	33.91	2.14
Ankle circumference (cm)	Males	225	19.1	25.3	22.23	1.36
	Females	228	18.2	25.1	21.40	1.36
Weight (kg)	Males	230	48.5	108.0	72.93	10.66
	Females	229	45.0	98.0	60.89	8.93
BMI (kg/m^2)	Males	229	17.2	37.1	23.96	3.16
	Females	229	16.3	40.6	23.13	3.50

Results

Descriptive statistics

The descriptive statistics including sex-specific means, ranges and standard deviations (SD) of the anthropometric measures, age and BMI are shown in Table 1. As can be seen, mean values for all anthropometric traits, as for the age, were higher in males than in females, confirming the already known sex mean dimorphism.

Factor analysis

Table 2 presents the results of the factor analysis after varimax rotation of the length and circumference measurements. In this analysis, the KMO (Kaiser-Mayer-Olkin value) was higher than 0.6 and the Bartlett´s test was significant ($p<0.001$), indicating a good adequacy of the sample to the analysis. The two components extracted in the factor analysis accounted for 76.77% of the total variance. LF explained 47.54% of the variance of the original traits and CF accounted for 29.23% of the total variance. These two components were used in the subsequent quantitative genetic analyses.

TABLE 2
FACTOR ANALYSIS WITH ORTHOGONAL VARIMAX ROTATION OF LENGTH AND CIRCUMFERENCE MEASUREMENTS

Traits	LF	CF
Height	0.957	0.169
Sternal height	0.947	0.171
Sitting height	0.800	0.262
Arm length	0.952	0.110
Upper arm length	0.863	–0.001
Forearm length	0.864	0.204
Middle finger length	0.798	0.213
Neck circumference	0.512	0.673
Upper arm circ. (rel.)	–0.076	0.911
Upper arm circ. (cont.)	0.005	0.921
Wrist circumference	0.566	0.642
Calf circumference	0.223	0.745
Ankle circumference	0.238	0.689
Eigenvalue	6.181	3.800
Percentage of total variance	47.54%	29.23%
KMO (Kaiser-Meyer-Olkin)	0.853	
Bartlett´s test	0.000	

LF – Linear factor, CF – Circumference factor

Univariate genetic analysis

Heritability estimates (h^2) with associated standard errors for the anthropometric traits and the two synthetic traits are given in Table 3. Also reported in this table is the significance of covariates (age, sex, age×sex, age^2, and age^2×sex). Sex was significant for all the variables, whereas the interaction between age and sex (age×sex) was not significant for most of the traits. All variables exhibit significant quadratic age trends except upper arm

TABLE 3
RESULTS OF UNIVARIATE QUANTITATIVE GENETIC ANALYSIS

Traits	Age	Sex	Age×sex	Age^2	Age^2×sex	Unadjusted h^2 of trait±SE	Adjusted h^2 of trait±SE[a]	Variance by covariates %
Height	***	***	n.s.	***	***	0.68±0.08	0.84±0.05	58
Sternal height	*	***	n.s.	***	**	0.77±0.07	0.87±0.05	53
Sitting height	**	***	n.s.	***	**	0.56±0.09	0.71±0.07	49
Arm length	n.s.	***	n.s.	*	*	0.74±0.08	0.81±0.06	51
Upper arm length	n.s.	***	n.s.	n.s.	n.s.	0.63±0.08	0.71±0.07	31
Forearm length	*	***	*	***	**	0.67±0.09	0.75±0.07	51
Middle finger length	n.s.	***	n.s.	*	*	0.86±0.07	0.88±0.05	46
LF	***	***	n.s.	*	*	0.70±0.08	0.86±0.06	57
Neck circumference	***	***	n.s.	***	**	0.45±0.13	0.67±0.07	67
Upper arm circ. (rel.)	***	***	n.s.	***	*	0.31±0.08	0.57±0.07	20
Upper arm circ. (cont.)	***	***	n.s.	***	*	0.29±0.08	0.55±0.07	22
Wrist circumference	***	***	n.s.	***	**	0.53±0.11	0.71±0.07	59
Calf circumference	n.s.	***	n.s.	***	*	0.54±0.09	0.56±0.09	15
Ankle circumference	n.s.	***	*	n.s.	n.s.	0.61±0.09	0.63±0.08	11
CF	***	***	n.s.	***	*	0.37±0.08	0.60±0.07	23
Weight	***	***	n.s.	***	***	0.44±0.10	0.65±0.07	39
BMI	***	**	n.s.	***	*	0.20±0.08	0.60±0.08	24

SE – Standard error, LF – Linear factor, CF – Circumference factor
n.s. not significant ($p>0.1$), *$p<0.1$, **$p<0.01$, ***$p<0.001$
[a] Heritability estimates are adjusted for significant covariates among age, sex, age × sex interaction, age^2, and age^2 × sex interaction.

length and ankle circumference. In general, adjustment for significant covariates increases the heritability estimates obtained for the studied traits. In fact, there is a strong influence of covariates on the variation of individual traits, collectively explaining 11–67% of the total variance of the traits. After accounting for the significant covariate effects, variance component analysis suggested that inter-individual differences in the studied variables were strongly dependent on additive genetic sources of variation. Heritabilities for all the studied traits were significant ($p<0.05$) and of substantial magnitude (>0.5). Middle finger length showed the highest heritability ($h^2=0.88$) and upper arm circumference (contracted) the lowest one ($h^2=0.55$). For the synthetic traits, the heritability estimates were 0.86 and 0.6 for LF and CF, respectively.

Bivariate genetic analysis

Additive genetic correlations (ρ_G), random environmental correlations (ρ_E), phenotypic correlations (ρ_P) and bivariate heritability (h^2_b) between pairs of anthropometric and synthetic traits (linear measurements and LF *vs*. height; circumference measurements and CF *vs*. BMI and weight; LF *vs*. height; CF *vs*. weight and BMI; and LF *vs*. CF) are presented in Table 4. The results demonstrated that both shared genes and common environmental factors contribute substantially to phenotypic covariation of these traits. All genetic correlations were significantly different from both 1.0 (data not shown) and zero (except the comparison between middle finger length and height), rejecting the hypothesis of complete pleiotropy and indicating incomplete pleiotropy (i.e. shared and unique sets of genes influenced these traits).

The additive genetic correlations (ρ_G) among linear measures and height were in general highly significant, after adjustment for significant covariates effects. The strength of the correlation ranged from moderate and non-significant (0.43) between middle finger length and height to a very high and significant estimate among sternal height and height (0.99). Common genetic factors (h^2_b) explained between 0.18 and 0.98 of the total residual variance of these traits. Concerning the comparisons made among circumference measures, all genetic correlations were significantly greater than zero ($p<0.001$)

TABLE 4
ADDITIVE GENETIC CORRELATIONS (ρ_G), ENVIRONMENTAL CORRELATIONS (ρ_E), PHENOTYPIC CORRELATIONS (ρ_P) AND BIVARIATE HERITABILITIES (h^2_b) BETWEEN ANTHROPOMETRIC MEASURES AND SYNTHETIC TRAITS ADJUSTED FOR TRAIT SPECIFIC COVARIATES

Comparisons	Genetic correlation (SE)	Environmental correlation	Phenotypic correlation	Bivariate heritability
Sternal height *vs.* Height	0.99 (0.00)***	0.81**	0.97	0.98
Sitting height *vs.* Height	0.85(0.04)***	0.30 n.s.	0.72	0.72
Arm length *vs.* Height	0.84(0.03)***	0.69**	0.82	0.71
Upper arm length *vs.* Height	0.81(0.05)***	0.31 n.s.	0.69	0.66
Forearm length *vs.* Height	0.70(0.05)***	0.54**	0.66	0.49
Middle finger length *vs.* Height	0.43(0.29)n.s.	0.50***	0.44	0.18
LF *vs*. Height	0.95(0.01)***	0.68*	0.91	0.90
Neck circumference *vs.* Weight	0.72(0.05)***	0.60***	0.68	0.52
Neck circumference *vs.* BMI	0.73(0.06)***	0.51***	0.65	0.53
Upper arm circ. (rel.) *vs.* Weight	0.72(0.05)***	0.71***	0.72	0.52
Upper arm circ. (rel.) *vs.* BMI	0.79(0.05)***	0.76***	0.78	0.62
Upper arm circ. (con.) *vs.* Weight	0.75(0.05)***	0.72***	0.74	0.57
Upper arm circ. (con.) *vs.* BMI	0.81(0.05)***	0.76***	0.79	0.65
Wrist circumference *vs.* Weight	0.69(0.06)***	0.18 n.s.	0.53	0.48
Wrist circumference *vs.* BMI	0.58(0.08)***	0.20 n.s.	0.44	0.33
Calf circumference *vs.* Weight	0.60(0.08)***	0.74***	0.65	0.36
Calf circumference *vs.* BMI	0.59(0.08)***	0.68***	0.63	0.35
Ankle circumference *vs.* Weight	0.66(0.07)***	0.42**	0.58	0.44
Ankle circumference *vs.* BMI	0.60(0.09)***	0.32*	0.49	0.36
CF *vs.* Weight	0.74(0.05)***	0.78***	0.75	0.55
CF *vs.* BMI	0.89(0.03)***	0.78***	0.85	0.79
LF *vs.* CF	–0.21(0.09)*	–0.60***	–0.29	0.05

SE – Standard error, LF – Linear factor, CF – Circumference factor, n.s. – not significant ($p>0.05$), * $p<0.05$, ** $p<0.01$, *** $p<0.001$

and of substantial magnitude (ρ_G=0.58–0.81), indicating that there are shared genetic effects influencing their expression. On the other hand, environmental correlations were also positive and significantly different from zero, except in the case of wrist circumference, indicating that these pairs of traits are influenced by shared environmental factors. The genetic correlation for each circumference measure vs. BMI and weight were of comparable magnitude. Between 0.36 and 0.57 of circumference measures variation was accounted for by genetic factors shared with body weight and between 0.33 and 0.65 with BMI. The negative genetic and environmental correlations between LF and CF implied that a set of genes and environmental factors with a positive influence on LF may exhibit negative effects on CF and vice versa.

Discussion

The variation of anthropometric phenotypes is due to a complex interaction between genetic and environmental determinants[12,32–34]. However, the complexity of these traits is not only caused by genetic and environmental factors, covariates such as sex and age also determined the variation of these phenotypes. In this study, as expected, sex and age differences were significant for all the traits. Although for some traits the main effect of age was not significant, the effects of age^2 or the interaction between age×sex or age^2× sex were significant for all the anthropometric traits except for upper arm length. Moreover, sex, age and their interactions account for 11 to 67% of the phenotypic variation of measured traits, supporting the evidence that the examination of the confounding effects of these two covariates constitute an important step for the analysis of the genetic structure of anthropometric phenotypes.

In order to ascertain the extent to which genetic factors influence the variation of body configuration related measures, we carried out a variance decomposition analysis which leaves no doubt regarding the substantial additive genetic effects on variation of these traits. Although contribution of genetic effects on body morphology have been investigated in some studies[18,19,21,35,36], comparison of results may be complicated since different variables were measured and different methods were used. Perhaps the most extensively studied body configuration measure is the BMI, which is considered a global index of corpulence and reflects the weight not only of fat tissue but also of muscle and skeletal tissues. Cross sectional twin and family studies have shown a moderate-to-substantial genetic component in its variation, being higher in twin designs (±70%) compared with family studies (±40%)[16]. The results of the present study suggest that BMI is significantly influenced by additive genetic factors, with narrow sense heritability estimate of 60%, which although is a high estimate is consistent with the results reported by other studies using family data[37,38].

Bearing in mind the heterogeneity of anthropometric measures, it is interesting to try to resume their variability in some independent factors which could provide a more general view of body configuration. In this study, LF can be clearly interpreted as a length factor. In relation to CF, bearing in mind the particular characteristic of some of the studied circumferences (e.g. ankle and wrist) that have been frequently used to characterize the frame size of the individuals and the high correlation coefficients obtained between this factor and two traditional indicators of body mass as are weight and BMI (0.74 and 0.89, respectively), we have interpreted this factor as an overall body mass factor. However, there remains the possibility that CF could be reflecting more soft tissue or even fat component variability rather than overall body mass because interindividual variation of circumferences in general population is mainly due to changes in fat component.

From this point of view, univariate variance component analysis showed that the variation of body length (LF) and body mass (CF), was strongly influenced by additive genetic effects (86% and 60%, respectively), which is in agreement with the study of Livshits et al.[18]. The strength of this heritability estimates suggests a higher importance of the genetic component determining length measurements than circumference measurements in this population, being these findings in line with the results of other studies[9,25,33,39–42].

Overall body morphology is composed of a set of different tissues or components, each of them presenting its own anatomical and physiological characteristics and also differing from the others, in the degree of genetic determination. Lengths are interpreted as skeletal dimensions because they are made between bony landmarks; however, the distances are also influenced by the soft tissues that overlie these bony landmarks. On the other hand, circumferences are composite measures determined by a combination of bone dimensions, muscle bulk, skin thickness and local body fat[43]. Thus, our findings support the previously known evidence of a greater genetic determination for measurements principally characterizing skeletal mass than in variables that also include soft-tissues (e.g. fat and muscle)[40–42,44,45]. In addition, a closer look at the patterning of circumferences shows that, variables more related to hard tissues such as wrist and ankle circumferences present higher values of heritability than soft tissue related measures (e.g. upper arm circumference and calf circumference), being these results in agreement with those of Devi and Reddi[41].

Afterwards, we investigate the possibility of shared genetic effects influencing covariation among pairs of anthropometric and synthetic traits (longitudinal measures and LF vs. height; circumference measures and CF *vs*. BMI and weight; LF *vs*. CF), since not only the variation of single traits, but also much of the covariation between different body configuration traits may depend on common genetic factors[18,19,21–46]. In addition to this, the high loadings of many variables on the same factor suggest that their variation is probably governed by the same genetic source (i.e. pleiotropy)[18]. Incomplete pleiotropy was detected between all pairs of traits except for middle finger length and height. It is remarkable that, al-

though these two measures did not show shared genes influencing their variation, both of them presented high heritability estimates, pointing to a great effect of additive genetic factors. However, it is clear that significant heritability estimates for two traits and a high ρ_P between them do not imply a significant ρ_G[31]. The lack of complete pleiotropy exhibited between the remaining traits indicates that, in addition to common genes with pleiotropic effect there is a unique set of genes controlling each anthropometric measure (2–67% of the genetic variance).

As well as for narrow sense heritabilities, higher genetic correlations were observed between length measurements vs. height (70–99%) than for circumference measurements vs. BMI or weight (58–81%). The highest genetic correlation was observed between sternal height and height; indicating that pleiotropic effects of a gene or a set of genes account for 98% of the genetic variance of the two traits. It is interesting to note that among comparisons between length measurements *vs*. height, measurements comprising a larger part of the body presented higher ρG values. Comparing with genetic correlations, environmental correlations are smaller in number and in the strength they correlate. Environmental correlations serve as a measure of the strength of the correlated response of two traits to nongenetic factors[47]. A noteworthy observation is that among length measurements, the comparison between sternal height and height presented the highest environmental correlation (81%) and sitting height *vs*. height the lowest one which may be explained by the fact that different nutritional environments during developmental stages modify more leg length than trunk length. In fact, leg length is a particularly sensitive indicator of childhood nutritional status and socioeconomic circumstances[48]. Among circumference measurements, upper arm circumferences relaxed and contracted vs. BMI showed the highest environmental correlation (76%) suggesting that environmental factors influencing one trait exert also their influence in the other trait.

In relation to the two synthetic traits, even though each of the factors extracted for the factor analysis are independent (i.e. varimax rotation) and therefore can not share common determinants, adjustment for the significant covariates allowed these common pleiotropic and environmental factors to be extracted[49]. Results for the current study clearly show that LF and CF share at least some common genetic and environmental effects (–21% and –60%, respectively) and the negative ρ_G and ρ_E implies that genes and environmental factors influencing LF have opposite effects on CF and *vice versa*. The human body growth in length and mass involves some common physiological processes, which may be a source of genetic correlation between them. Some regulation factors control these common physiological processes, consequently all body measurements may share some genes contributing to these phenotypic variables. In light of these results, it seems that even the variation of body linearity and body mass is highly determined by genetic factors, showing heritability estimates higher than 50% (86% and 60%, respectively), the shared genetic influences are unlikely to explain much of the observed variation between the two factors (5%). Thus, while univariate heritabilities indicated that LF and CF have a great genetic component influencing their expression, the influence of shared genes on these phenotypes does not substantially contribute to their expression being more determined by different subsets of genes.

Several limitations of the present study should be considered: As previously mentioned, heritability data for simple anthropometric traits were already estimated in Susanne[9] but we have recalculated them because univariate analysis is a necessary step previous to bivariate analysis, in which the significant covariates are defined. In addition to this, the variance components analysis improves the accuracy of heritability estimates compared to regression model employed by Susanne[9]. On the other hand, maximum likelihood techniques can be sensitive to small sample sizes and the power of variance components test increased significantly with increases sample size, therefore another potential limitation of the present study could be the number of studied nuclear families. Nevertheless, the results of variance component analyses have been significant and therefore provide valid and robust information about the genetic and environmental contribution to variation in body configuration in Belgian nuclear families.

Although there is a wealth of publications analysing the involvement of genetic factors in the variation of body configuration traits, little is known about shared genetic and environmental effects determining body morphology. To our knowledge, this is the first study in which the shared genetic and environmental factors influencing body lengths and circumferences. Thus, further studies might need to be performed in different populations to disentangle the genetic interactions underlying the genetic and environmental determination of these traits' variability. These results could also be very useful in the search for genes generating the genetic variance of these anthropometric measures (e.g. quantitative traits linkage analysis), which constitutes an important challenge for future research.

Conclusion

In conclusion, our data provide reliable evidence for the substantial role of genetic factors in the determination of the phenotypic variability of body configuration in this Belgian population. Narrow-sense heritability estimates indicate a greater genetic determination for measurements principally characterizing skeletal mass than in variables that also involve soft-tissues. Bivariate decomposition analyses show that pleiotropic effects have a great influence in determining body traits variation among body linear measurements, as well as between body circumferences and weight or BMI. Environmental factors have less influence than pleiotropy on all the analysed traits. Finally, in relation to overall body configuration, even though genetic and environmental control of hu-

man body length and mass is mainly determined by different subset of genes a little but significant part of body configuration is due to shared genetic and environmental effects.

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GENETIČKI DOPRINOS VARIJACIJI S OBZIROM NA VELIČINU TIJELA I TJELESNU MASU KOD BELGIJSKIH NUKLEARNIH OBITELJI

SAŽETAK

Cilj ove studije bilo je procijeniti doprinos genetičkih faktora na fenotip povezan s konfiguracijom tijela. Uzorak se sastojao od 119 belgijskih nuklearnih obitelji i uključivao je 231 muškarca i 229 žena. Faktorska analiza je provedena kako bi se analiziralo 13 mjera dužine i opsega, a dobivene sintetičke karakteristike (LF i CF, linerni i opsežni faktori) su iskorišteni kao varijable. Univarijatna kvantitativna genetička analiza pokazala je da i antropometrijske i sintetičke karakteristike značajno ovise o utjecaju genetike, s faktorom nasljednosti u rasponu od 0.55 do 0.88. Procjene nasljednosti su bile više za mjere koje su karakterizirale koštanu masu, za razliku od onih za mekano tkivo. Spol, dob i

njihova međusobna interakcija objašnjavale su 11–67% ukupne fenotipske varijance. Također je ispitana i kovarijacija između parova antropometričkih i sintetičkih karakteristika (mjere dužine i LF *vs.* visina; mjere opsega i CF *vs.* težina i indeks tjelesne mase; LF *vs.* CF). Značajna genetička korelacija između svih ispitivanih karakteristika (osim za dužinu *vs.* visinu srednjeg prsta ruke) potvrdila je utjecaj pleiotropije na genetičku uvjetovanost ovih karakteristika. Bivarijatna analiza pokazala je da pleiotropija ima velik utjecaj pri utvrđivanju varijacije tjelesnih karakteristika kod mjerenja tjelesne dužine, kao i kod tjelesnog opsega, težine i indeksa tjelesne mase. S obzirom na dvije sintetičke karakteristike, čak su i varijacije tjelesnih dužina i opsega bili pod snažnim utjecajem genetičkih faktora. Dijeljeni genetički faktori nisu uspjeli objasniti opaženu varijaciju između LF i CF. Rezultati ove studije nam omogućuju da zaključimo kako su u ovoj populaciji karakteristike povezane s tjelesnom konfiguracijom pod snažnim utjecajem genetike te da dijeljeni geni također doprinose toj genetičkoj strukturi.