Inheritance of six anthropometric traits in Vaidyas of West Bengal, India

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Summary

Background: Anthropometric traits are important quantitative traits used by biological anthropologists. Surprisingly little is known about their pattern of inter-generational transmission, probably due to lack of use of developed statistical analysis in previous studies.

Aim: The present study is an attempt to approach the problem of the inheritance of anthropometric traits through advanced statistical applications.

Subjects and methods: Measurements of 824 individuals from 200 families including two generations were collected from Barasat, West Bengal. The study includes age correction by regression, familial correlation, heritability estimation and segregation analyses.

Results: Results showed that there is strong involvement of the familial component in variation of anthropometric traits. The magnitude of heritability ($h^2 = 57$–83%) also supports their strong genetic basis. The results indicated that additive genes are not the only contributing factor; the effect of environment is considerable and that of dominant genes not negligible. Due to genetic interaction with the local environment (specific for each trait), heritabilities vary from one trait to another. Length measurements have higher heritability than breadth measurements. Segregation analysis revealed that either the additive or dominant major gene (MG) is responsible for this effect, which follows simple Mendelian transmission. Beside this, the possibility of the existence of an additional minor gene cannot be discarded.

Conclusion: Anthropometric traits have a genetic basis but their mode of inheritance is quite complex in nature. There is evidence of major gene effect (along with polygenes) with Mendelian transmission.

Keywords: *Inheritance, anthropometry, endogamous population, India*

Introduction

Anthropometry is a major tool 'for studying mankind through time and space' (Sen 1994). Unfortunately the mode of inheritance of different anthropometric traits has not been clearly established. Growth studies, particularly longitudinal ones, have produced valuable knowledge on genetic control, but these previous longitudinal studies have usually been confined to height and weight (Roberts 1981; Livshits et al. 2000; Pietilainen et al. 2002).

Most inheritance studies have also been conducted only on height, weight and body mass index (BMI) (Byard et al. 1985; Dasgupta et al. 1997; Ginsburg et al. 1998; Colilla et al. 2000; Raychaudhuri et al. 2003; Škarić-Jurić et al. 2003). Relatively few studies have focused on the pattern of inheritance of the whole range of anthropometric traits (Mueller and Titcomb 1977; Poosha et al. 1984; Sharma 1986; Livshits et al. 2002; Ermakov et al. 2005). Studies of chromosomal abnormalities and other single gene disorders are no less valuable as they reveal how genes control postnatal growth (Roberts 1981). On the other hand, embryological studies have revealed the mechanisms of growth of structures highlighting possible sources of environmental influence and the role of genetic factors (Sperber 1981; Ijzerman et al. 2001; Schwarzler et al. 2003; Power et al. 2003). However, these studies have not succeeded in throwing light on the mechanism of genetic control of these quantitative traits.

A review of the literature revealed that studies of the inheritance pattern of anthropometric traits are very few (especially in India) and information on the contribution of genetic factors to these traits is inadequate. This is probably due to the fact that the genetics of these quantitative traits is very complex in nature, requiring modern techniques of statistical analysis. Although genome research has become a popular and powerful technique for identifying candidate-gene regions of quantitative traits, this technique of genetic analysis is very expensive for a developing country like India, and blood samples are not available in many instances. In these cases, segregation analysis can substantially advance our knowledge of anthropogenetics. However, most previous inheritance studies were simply based on the application of correlation or regression between relatives. To our knowledge, until now hardly any studies are available using a genetic model test on the whole range of anthropometric traits from different parts of the body. Thus, there is scope for more investigation in this field. Therefore, the aim of the present study is to report the magnitude and mode of inheritance of some anthropometric traits on an endogamous population through segregation analysis along with some traditional analyses like correlation, regression or heritability estimation.

Materials and methods

The present study has been confined to an endogamous caste, Vaidyas (for a detailed description see Sengupta and Karmakar 2003), from Barasat in the district of North 24-Parganas of West Bengal. The data consists of 824 individuals (male: 429, female: 395) from 200 families comprising living parents (father: 200, mother: 200) with at least two children (son: 229, daughter: 195). The geographical location of the studied area (a subtropical area with a monsoon regime) is presented in Figure 1. The exact time and origin of this caste as a separate caste group is disputed by scholars. Some consider the Vaidyas to be an offshoot of the Brahmins who intermarried with other castes like Vaishya, Sudra, etc., but there are also claims that they are direct descendents of Aryans who immigrated to Bengal (Dutta 1969). They generally occupy a high rank in society and are traditionally recognized as physicians. Today most of them are engaged in white-collar jobs.

Some basic information about the subject(s) was taken, including name, age and sex. As most of the individuals in the sample were well educated, accurate age was recorded with certainty. The age of the first generation ranges between 40 and 72 years and for their children from 12 to 34 years. Care was taken to ascertain that the samples/individuals were physically normal with no apparent defects either in the limbs or in the face and head (micromegaly, macromegaly, Down's syndrome, etc.).

Figure 1. Map of West Bengal showing the geographical location of the studied population in 24-Parganas.

As it is also interesting to know whether there is any difference between length and breadth measurements with respect to inheritance pattern, the present study considered six length and breadth measurements from three different areas (head, upper and lower extremities) of the body. Using a Martin's slide caliper, measurements were collected (Lohman et al. 1988) from both sides of the body, and the average values were recorded (these measurements will be analysed in another study to investigate the inheritance of bilateral asymmetry). Details of the measurements are given in the first column of Table I. All the measurements were taken by the same investigator (the first author) to minimize inter-investigator error.

Statistical analysis

The basic descriptive statistics were carried out including mean, standard deviation (SD) and standard error (SE). Because of the age effect on anthropometric traits and as the studied individuals were of different ages, the possible dependent variables were adjusted for age by regression analysis on age, age^2, and age^3. These standardized residuals are the phenotype variables used for further analysis.

The degree of resemblance was studied by familial correlation following two methods: (a) interclass—between parents and their children as well as the parents themselves (by Karl Pearson), (b) intraclass—between sibs, which means brothers and sisters (by Fisher 1958).

Table I. Descriptive statistics of anthropometric traits.

Variables	Father ($n=200$)		Mother ($n=200$)		Son ($n=187\star$)		Daughter ($n=148\star$)	
	Mean $\pm$ SE	SD	Mean $\pm$ SE	SD	Mean $\pm$ SE	SD	Mean $\pm$ SE	SD
Ear length	6.71 ± 0.04	0.50	6.23 ± 0.03	0.40	6.83 ± 0.13	0.43	6.31 ± 0.03	0.35
Ear breadth	2.95 ± 0.02	0.26	2.71 ± 0.02	0.25	2.89 ± 0.02	0.16	2.66 ± 0.01	0.10
Hand length	17.83 ± 0.06	0.81	16.61 ± 0.06	0.80	17.71 ± 0.09	1.02	16.72 ± 0.10	1.21
Hand breadth	7.82 ± 0.03	0.36	7.18 ± 0.02	0.34	7.91 ± 0.05	0.73	7.04 ± 0.04	0.65
Foot length	24.11 ± 0.08	1.20	22.33 ± 0.08	1.12	23.99 ± 0.14	2.41	22.11 ± 0.13	1.32
Foot breadth	9.34 ± 0.04	0.55	8.48 ± 0.04	0.58	9.38 ± 0.04	0.75	8.32 ± 0.05	0.69

* Only children over 18 years are included here.

The heritability of these traits was calculated by Falconer (1960).

Complex segregation analysis was carried out using Maximum Likelihood Methods by the 'Pedigree Analysis Package' (PAP) (Hasstedt 1994), to evaluate the mode of quantitative inheritance. The program estimates the allele frequency (p), transmission probabilities (τ_{A1A1}, τ_{A1A2} and τ_{A2A2}), three genotypic means (μ_{A1A1}, μ_{A1A2} and μ_{A2A2}) with their standard deviations (σ_{A1A1}, σ_{A1A2}, σ_{A2A2}) and heritability of the traits (h^2). Seven genetic models—(1) general model, (2) sporadic model, (3) environmental model, (4) τs equal to p model, (5) Mendelian model, (6) no polygenic component model, and (7) most parsimonious Mendelian (MP) model—were tested. These models were described in detail in our previous papers (Sengupta and Karmakar 2004). Models 2–6 are the sub-models of the general model and thus $-2\ln L$ (log likelihood) values of these models were compared to that of the general model in order to choose the best one. Hypothesis 7 is the sub-models of the Mendelian model (the fifth one) and, therefore, was tested against it.

The major gene (MG) variance and total variance was calculated from the MP model using the same package (Hasstedt 1994). No ascertainment correction of likelihood was made because our method of pedigree collection was unrelated to the anthropometric traits of the pedigree members.

Results

The overall descriptive statistics (generations and sex separately) including mean standard deviation (SD) of the studied traits are presented in Table I. As in the offspring generation some individuals were under 18 years, only children of more than 18 years were considered for comparisons to parents (for the same reason, the values are corrected for age before going to further analyses). Length measurements were, in general, higher than their respective breadth measurements, and among length measurements, foot length was found to be the highest value. For both sexes, mean values of offspring generations were not significantly different from their parental generations.

The results of inter- and intra-class correlation values are presented in Table II. The variables exhibited very low (even some are negative) non-significant husband–wife correlation. For other relationships, with an exception of brother–brother correlation of ear breadth ($r=0.15$), all the other values are statistically significant ($p<0.01$ or $p<0.05$). Ear length showed the highest correlation value for parent–child ($r=0.39$) and sib–sib ($r=0.45$) combination, respectively, while midparent–child correlation was highest in both ear and foot length ($r=0.50$). There was no greater difference among four types

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Table II. Familial correlation of anthropometric traits.

Variables	HW	FS	FD	MS	MD	FC	MC	PC	MidS	MidD	MidC	BB	BS	SS	Sib	h^2
n	200	229	195	229	195	424	424	848	229	195	424	152	240	110	502	
Ear length	0.04	0.48**	0.36**	0.36**	0.37**	0.43**	0.37**	0.39**	0.59**	0.41**	0.50**	0.46**	0.47**	0.43**	0.45**	0.83
Ear breadth	0.06	0.28**	0.31**	0.30**	0.26**	0.30**	0.28**	0.26**	0.36**	0.35**	0.36**	0.15	0.35**	0.30**	0.27**	0.57
Hand length	−0.03	0.42**	0.32**	0.28**	0.36**	0.38**	0.32**	0.34**	0.49**	0.49**	0.49**	0.47**	0.44**	0.39**	0.38**	0.74
Hand breadth	−0.02	0.30**	0.36**	0.24**	0.32**	0.33**	0.28**	0.31**	0.39**	0.47**	0.43**	0.45**	0.42**	0.46**	0.41**	0.64
Foot length	0.03	0.40**	0.31**	0.33**	0.38**	0.36**	0.35**	0.36**	0.50**	0.50**	0.50**	0.43**	0.44**	0.34**	0.41**	0.77
Foot breadth	0.04	0.40**	0.27**	0.17**	0.38**	0.34**	0.37**	0.31**	0.40**	0.44**	0.42**	0.32**	0.46**	0.47**	0.40**	0.63

n, number of pairs; $*p < 0.05$; $**p < 0.01$; HW, husband–wife; FS, father–son; FD, father–daughter; MS, mother–son; MD, mother–daughter; FC, father–child; MC, mother–child; PC, parent–child; MidS, midparent–son; MidD, midparent–daughter; MidC, midparent–child; BB, brother–brother; BS, brother–sister; SS, sister–sister; Sib, sib–sib.

of parent–offspring combinations. Mother–child correlation was not higher than father–child correlation in any of the studied traits. Except foot length and foot breadth, brother–sister ($r = 0.44$ and $r = 0.46$, respectively) correlation was lower than brother–brother ($r = 0.43$ and $r = 0.32$, respectively). Without exception, parent–child correlation was lower than midparent–child and sib–sib correlation.

The magnitudes of heritability estimates are presented in the last column of Table II. All of the anthropometric traits have heritability of more than 50% (varying from 57 to 83%). The length measurements have higher heritability (more than 0.70) than the breadth measurements. Ear length showed the highest heritability ($h^2 = 0.83$) and physiognomic ear breadth the least ($h^2 = 0.57$).

The segregation analysis for the studied traits is given in Table III. The table presents maximum likelihood estimates of the model parameters, $-2\ln L$ values and respective χ^2 values with their degrees of freedom. Model 2 or the sporadic model, which assumes no inter-generational transmission, was tested first and rejected at 1% level. The environmental model (model 3) was also not accepted when it was compared with the general model ($\chi^2 > 5.99$, d.f. $= 2$, $p < 0.05$). Both model 4 (τs equal to p), which assumes the non-transmission of the major gene, and model 6 (or the model of no-polygenic component) were rejected (either $p < 0.05$ or $p < 0.01$). The only model that was accepted for all anthropometric traits was the Mendelian model ($\chi^2 < 7.81$, d.f. $= 3$, $p > 0.05$). As a next step, the MP model was tested to evaluate the mode of major effect on these traits. The Mendelian model was constrained with dominant, additive and recessive sub-models. It was found that ear length was compatible with the dominant model, while other traits followed the additive model. Among all these models, only the general model and best fitting MP model were present for all traits.

The total genetic variance and MG variance of the anthropometric traits obtained from the MP model are presented in the last two rows of Table IV. The table shows that 26–35% of the total variation was attributable to genetic effects. The putative major gene identified by this model accounted for only 7–10% of total phenotypic variance in these traits.

Discussion

Body size and shape seem to be the product of the continuous and complex interaction of genetical (G) and environmental (E) factors. This interaction is non-linear, i.e. the relationship cannot be shown by a plus sign ($G + E$), rather a product of them ($G \times E$). As local environment is not same for all parts of the body, the present study includes the inheritance of anthropometric measurements from three major areas of the body like head, upper and lower extremities.

All traits, irrespective of area, showed a weak and non-significant association between husband and wife, indicating negligible assortative mating for these traits, which coincides with previous studies on body size (Susanne 1979; Kaur and Singh 1981) and shape (Sánchez-Andrés 1995). The magnitude of correlation between other pair relationships suggests a strong involvement of genetic components, which is in agreement with limited earlier studies on this topic (Susanne 1975; Sharma et al. 1984; Kapoor et al. 1985). The correlation values of the present study are slightly lower than the theoretical value suggested by Fisher (1918) and Penrose (1949), which may indicate the involvement of an environmental effect also. Higher correlations between sibs than between parent and offspring also support the presence of environmental factors, because environmental factors make a more important contribution to the correlation between sibs than between parent

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Table III. Segregation analysis of anthropometric traits.

Parameter	Ear length		Ear breadth		Hand length		Hand breadth		Foot length		Foot breadth	
	General model	MP model	General model	MP model	General model	MP model	General model	MP model	General model	MP model	General model	MP model
ρ	0.526	0.584 ± 0.009	0.517	0.336 ± 0.014	0.571	0.180 ± 0.005	0.532	0.393 ± 0.007	0.682	0.631 ± 0.008	0.512	0.582 ± 0.008
τ_1	0.893	1.00*	0.969	1.00*	0.932	1.00*	0.915	1.00*	0.947	1.00*	0.995	1.00*
τ_2	0.477	0.50*	0.412	0.50*	0.556	0.50*	0.428	0.50*	0.318	0.50*	0.496	0.50*
τ_3	0.182	0.00*	0.014	0.00*	0.182	0.00*	0.155	0.00*	0.024	0.00*	0.140	0.00*
μ_1	0.553	0.454 ± 0.014	−0.521	0.412 ± 0.035	0.512	0.580 ± 0.010	0.592	0.282 ± 0.010	0.382	0.171 ± 0.012	0.344	0.182 ± 0.010
μ_2	0.936	0.454††	−0.126	0.153§	−0.016	0.283§	0.604	0.051§	0.559	0.401§	0.512	0.422§
μ_3	−0.254	0.034 ± 0.016	0.569	0.106 ± 0.009	−0.529	-0.015 ± 0.004	0.447	-0.018 ± 0.014	0.647	0.631 ± 0.006	0.721	0.662 ± 0.014
σ_1	0.726	0.521 ± 0.018	0.682	0.642 ± 0.011	0.712	0.482 ± 0.006	0.617	0.582 ± 0.008	0.621	0.515 ± 0.010	0.632	0.517 ± 0.007
σ_2	0.593	$0.608 \pm 0.0.011$	0.713	0.458 ± 0.008	0.669	0.691 ± 0.012	0.580	0.439 ± 0.006	0.517	0.641 ± 0.014	0.754	0.463 ± 0.006
σ_3	0.668	0.446 ± 0.015	0.558	0.532 ± 0.014	0.593	0.515 ± 0.009	0.714	0.721 ± 0.010	0.782	0.407 ± 0.008	0.556	0.507 ± 0.012
h^2	0.810	0.510 ± 0.009	0.609	0.567 ± 0.015	0.763	0.583 ± 0.010	0.695	0.315 ± 0.008	0.668	0.472 ± 0.006	0.371	0.411 ± 0.011
$-2\ln L$	2176.264	2184.255	2053.624	2058.486	2041.258	2043.276	2142.625	2146.354	2085.478	2089.630	2156.5447	2264.822
χ^2	–	1.17 (1)	–	0.657 (1)	–	1.497 (1)	–	1.624 (1)	–	2.641 (1)	–	2.333 (1)

(n), Number of degrees of freedom.
*Parameter is fixed at given value. ††Parameter constrained to equal parameter value listed above it. ‡Model is dominant. §Model is additive.

Table IV. Total variance and MG variance of anthropometric traits.

Serial no.	Variables	Total variance	MG variance
1.	Ear length	35	8
2.	Ear breadth	29	10
3.	Hand length	35	8
4.	Hand breadth	33	7
5.	Foot length	26	10
6.	Foot breadth	32	9

and child (Matsuda 1973; Susanne 1975). Neither the measurements of the head nor of two extremities followed the hypothesis of involvement of the X-linked gene (Hewitt 1957). Non-significant difference found in the dependence of children's measurements on those of their fathers and mothers taken separately suggests low maternal effect of these variables.

The heritabilities of these morphological characters varied from 51 to 77%. Livshits et al. (2002) also showed that 40–75% of the inter-individual variation of body size and shape was attributable to genetic factors. But the magnitude of heritability in the present study varies from one trait to another, which may be due to genetic interaction with the local environment specific for each trait. Ear length showed the highest heritability coefficient ($h^2 = 0.77$), which was also observed ($h^2 = 0.81$) in a Punjabi population of India by Sharma (1986). For both head and extremities, the heritabilities of length measurements in the present study are higher than those of breadth measurements, which is in line with the results of other studies (Tanner and Israelson 1963; Susanne 1975; Kaur and Singh 1981; Sánchez-Andrés and Mesa 1994). The reason for the higher contribution of genetic factors to length measurements may be described from the nature of their growth. A bone grows in length by ossification of the diaphyseal surface of the cartilage, but in width by new layers of bone (a sheet of fibrous materials called periosteum) being deposited on it (Wilson and Waugh 1996). Thus, growth of breadth may be more plastic than length in development responding considerably to stress and strains.

The higher heritability of length measurements of the head than the measurements of extremities may be due to differential growth of the body proportions of the foetus. According to Malina and Bouchard (1991), the tissues associated with brain, nervous system and associated structures (i.e. eyes, upper face, parts of skull, etc.) experience rapid growth. At 2 months of pregnancy, the developing foetus has an enormous head relative to overall size and short extremities. This differential growth of body proportion occurs until mid-pregnancy, after which growth occurs in the trunk and extremities. Thus the head is exposed to the intra-uterine environment for less time than the other parts of the body such as the extremities, leading to higher heritability of this part.

The variation in familial resemblance and heritability between measurements of upper and lower extremities can also be explained from their time of appearance and growth during prenatal life. At 32–34 days old, the paddle-shaped fore and hind limbs of the human embryo make their appearance. The forelimbs appear earlier and subdivide into arm, forearm and hand in a 40-day-old embryo, while the hind limbs still retain their primitive shape. The digits of the hand are clearly separated within 46 days, but not the foot (Hamilton et al. 1963). Furthermore, not all genes are active at the same time in both prenatal and postnatal life. At each stage of development some genes are turned on and other turned off, and the molecules that regulate the action may either be resident in the cell or may diffuse out of the cells in which they are produced and regulate neighbouring cells.

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Therefore, they may exert differential actions on cells at different distances, and different positions of the body of the embryo (Tanner 1978) causing differential growth on these areas.

However, the evidence for the existence of a putative major gene (MG) on the studied traits was investigated by segregation analysis. The result of χ^2 showed that the model denying the transmission of genetic effect (sporadic model) was strongly rejected for all traits, indicating the possibility that the effect is transmitted in the families. Taking this into account, other models were tested and the environmental models with equal τs and no MG model were rejected, suggesting the possibility of the influence of MG on these traits. When the Mendelian model was tested, the pattern of genetic transmission was consistent with a MG effect. But the rejection of the no polygenic component model does not rule out the possibility of the existence of additional minor genes also (polygenes). The result of the MP model, which is used to understand the mode of Mendelian transmission, showed that only the additive hypothesis was not rejected ($p > 0.05$) and provided the best fit for all traits except ear length, which accepted the dominant model.

Although the MG model was accepted for these anthropometric traits, the evidence for MG from different populations makes it possible to consider this model as a phenomenon influencing body size. But studies concerned with segregation analysis of different parts of body measurements are scarce. Although the influence of MG effects on the studied anthropometric traits is not available in the literature, this effect has been already reported in other anthropometric variables such as height, weight and BMI (Bouchard et al. 1998; Ginsburg et al. 1998; Škarić-Jurić et al. 2003). Li et al. (2004) suggested that MG with a recessive effect accounts for about 17.2% of the total adult height variation in the Chinese. Rice et al. (1993) found that body mass assessed by underwater technique and per cent of body fat are satisfactorily described by the MG model. Comuzzie et al. (1995) also reported that a mixed Mendelian model could be accepted for fat mass. Evidence for a MG influence on visceral area fat was also reported from different studies (Bouchard et al. 1996; Rice et al. 1997). Livshits et al. (1995) showed that MG is involved in the determination of human body size as assessed by means of principal component analysis of the large array of morphological traits, although polygenes were also found to contribute to the total variation of these variables. In the same way, if the MG model is accepted in the studied traits from ethnically and geographically different populations, then the existence of this model can be taken as a reliable result.

Acknowledgements

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