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Apportionment of Global Human Genetic Diversity Based on Craniometrics and Skin Color

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ABSTRACT A number of analyses of classical genetic markers and DNA polymorphisms have shown that the majority of human genetic diversity exists within local populations (≈85%), with much less among local populations (≈5%) or between major geographic regions or "races" (≈10%). Previous analysis of craniometric variation (Relethford [1994] Am J Phys Anthropol 95:53–62) found that between 11–14% of global diversity exists among geographic regions, with the remaining diversity existing within regions. The methods used in this earlier paper are extended to a hierarchical partitioning of genetic diversity in quantitative traits, allowing for assessment of diversity among regions, among local populations within regions, and within local populations. These methods are applied to global data on craniometric variation (57 traits) and skin color. Multivariate analysis of craniometric variation shows results similar to those obtained from genetic markers and DNA polymorphisms: roughly 13% of the total diversity is among regions, 6% among local populations within regions, and 81% within local populations. This distribution is concordant with neutral genetic markers. Skin color shows the opposite pattern, with 88% of total variation among regions, 3% among local populations within regions, and 9% within local populations, a pattern shaped by natural selection. The apportionment of genetic diversity in skin color is atypical, and cannot be used for purposes of classification. If racial groups are based on skin color, it appears unlikely that other genetic and quantitative traits will show the same patterns of variation. Am J Phys Anthropol 118:393–398, 2002.

A major finding of genetic studies of human populations is the demonstration that the majority of human genetic diversity exists within local populations, with much less diversity among local populations or among geographic regions, or "races" (Brown and Armelagos, 2001). In his pioneering study, Lewontin (1972) analyzed worldwide variation in allele frequencies for a number of genetic markers, and found that variation among major geographic regions accounts for 6.3% of total variation, and that variation among local populations within geographic regions accounts for 8.3% of the total variation. The remaining 85.4% of genetic variation exists within local populations. Additional studies based on classical genetic markers, primarily blood groups and blood protein polymorphisms (Latter, 1980; Ryman et al., 1983) and DNA polymorphisms (Barbujani et al., 1997; Jorde et al., 2000), found similar results. These studies reflect neutrality on average, and show that variation among geographic regions accounts for roughly 10% of the total genetic diversity in our species, while 5% is due to variation among local populations within regions, and 85% within local populations.

Less attention has been given to the apportionment of genetic diversity in quantitative traits. Some have suggested that among-region differences in many morphological and metric traits are greater than for genetic markers (e.g., Nei and Roychoudhury, 1982; Stringer and Andrews, 1988), perhaps because of the effect of natural selection on increasing geographic differentiation. In an earlier paper (Relethford, 1994), I showed that between 11–14% of our species' craniometric diversity occurs among geographic regions, with the remaining 86–89% occurring within regions.

The purpose of this paper was to extend that analysis with a hierarchical design that considers three sources of variation in a quantitative trait: 1) variation among geographic regions, 2) variation among local populations within geographic regions, and 3) variation within local populations. This method is applied to global data on craniometric variation and skin color. The results show that the components of variation for craniometric traits closely resemble those obtained from genetic marker and DNA polymorphism data, whereas the components of varia-

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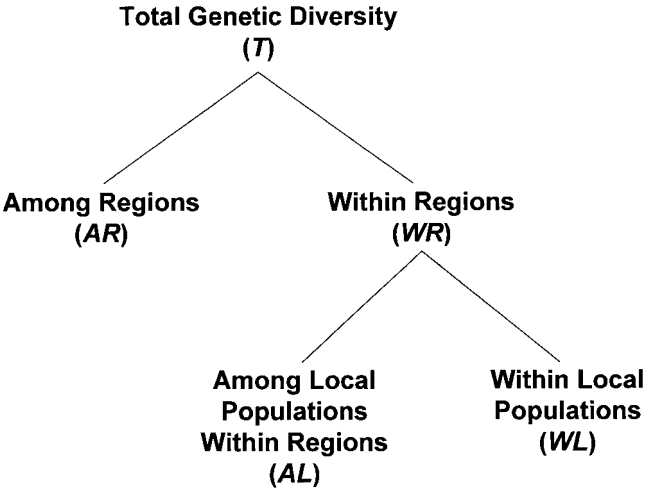

Fig. 1. Hierarchical partitioning of global genetic diversity.

tion in skin color are quite different, reflecting a geographic pattern of past natural selection.

APPORTIONMENT OF QUANTITATIVE VARIATION

Following Lewontin (1972) and others, I consider a hierarchical partitioning of global genetic diversity. The total species (T) is subdivided into a number of geographic regions (R), each of which is further subdivided into a number of local populations (L). Three components are of interest:

AR, the proportion of total diversity among geographic regions;
AL, the proportion of total diversity among local populations within geographic regions; and
WL, the proportion of total diversity within local populations.

The partitioning is hierarchical because the proportion of total genetic diversity within regions (WR) is the sum of the nested components: WR = 1 − AR = AL + WL (see Fig. 1).

Genetic diversity is often assessed from allele frequencies using the average per-locus heterozygosity, computed as

$$H = 1 - \frac{\sum_k p_k^2}{n} \tag{1}$$

where n is the number of loci, k is the total number of alleles over all n loci, and summation is over all alleles and loci (Nei, 1987). Following Lewontin (1972) and others, partitioning of diversity is done by comparing average per-locus heterozygosities at three levels of analysis: 1) total heterozygosity (H_T) obtained by pooling data from all populations in all regions, 2) regional heterozygosity (H_R) obtained by computing heterozygosity for each region separately and averaging these values, and 3) local heterozygosity (H_L) obtained by computing heterozygosity separately for each local population and averaging these values. The variance components are then computed as

$$\begin{aligned} AR &= \frac{H_T - H_R}{H_T} \\ AL &= \frac{H_R - H_L}{H_T} \\ WL &= \frac{H_L}{H_T} \end{aligned} \tag{2}$$

Note that $AR + AL + WL = \frac{H_T}{H_T} = 1$.

Because the genotypic variance of a quantitative trait is proportional to heterozygosity under an equal and additive effects model of quantitative inheritance (Falconer, 1981), partitioning of the variance components using quantitative traits seems an easy task. However, the genotypic variance equivalent to total heterozygosity cannot directly be observed, and is *not* the same as the genotypic variance obtained by pooling all individuals, a quantity that includes both within-group and among-group variance components (Rogers and Harpending, 1983; Relethford and Blangero, 1990). In order to estimate variance components from quantitative traits, I use the relationship between heterozygosity and Wright's F_{ST}, a measure of reduction in heterozygosity due to the subdivision of a total population (T) into a number of subpopulations (S), where

$$F_{ST} = \frac{H_T - H_S}{H_T} \tag{3}$$

This formulation is useful because F_{ST} can be estimated from quantitative traits (for computational methods, see Williams-Blangero and Blangero, 1989; Relethford and Blangero, 1990; Relethford et al., 1997).

F_{ST} can be estimated for different levels of population aggregation and then used to estimate variance components. A given data set is analyzed at two levels, first by geographic region (R) and then by local population (L). The first F_{ST} compares variation among regions relative to total variation

$$F_{RT} = \frac{H_T - H_R}{H_T} \tag{4}$$

The second F_{ST} compares variation among local populations relative to total variation

$$F_{LT} = \frac{H_T - H_L}{H_T} \tag{5}$$

Combining Equations (2), (4), and (5) allows the variance components to be estimated from the observed values of F_{RT} (the F_{ST} obtained when using regions as the unit of analysis) and F_{LT} (the F_{ST} obtained when using local populations as the unit of analysis) as

$$AR = F_{RT}$$

TABLE 1. Regional sample sizes for craniometrics and skin color

Data set	Geographic region	Sample size	Number of local populations
Craniometrics	Sub-Saharan Africa	283	3
	Europe	317	3
	East Asia	261	3
	Australasia	298	3
	Polynesia	294	3
	Americas	281	3
	Total	1,734	18
Skin color	Sub-Saharan Africa	2,023	27
	Europe	2,684	22
	Central/East Asia	675	8
	Australasia	659	7
	Americas	225	4
	Total	6,266	68

$$\begin{aligned} WR &= 1 - F_{RT} \\ AL &= F_{LT} - F_{RT} \\ WL &= 1 - F_{LT} \end{aligned} \quad (6)$$

MATERIALS AND METHODS

The above methods were applied to global data sets representing craniometric and skin color variation within and among human populations. The craniometric data were originally collected by Howells (1989) and consist of 57 craniometric measurements on 1,734 crania from 18 local populations in six major geographic regions: Sub-Saharan Africa, Europe, East Asia, Australasia, Polynesia, and the Americas. Regional sample sizes are reported in Table 1. Each region consists of three local populations. Lists of specific variables and populations are given by Howells (1989) and my earlier analyses of this data set (Relethford, 1994; Relethford and Harpending, 1994). Following this earlier work, data on males (N = 907) and females (N = 827) were pooled after converting all variables to standardized scores within each sex.

Skin color variation was assessed using published means and variances for skin reflectance measured on the E.E.L. portable reflectance spectrophotometer using filter 609, which samples the visible wavelength at 685 nm and is the best single index of skin color. Only data from males are used here, because the published literature includes summary statistics for more male samples than female samples. The total compiled database was described elsewhere (Relethford, 1997, 2000). A portion of these data is used here, consisting of the summary statistics based on 6,266 individuals in five major geographic regions: Sub-Saharan Africa, Europe, Central/Eastern Asia, Australasia, and the Americas. Regional sample sizes are reported in Table 1. These data were chosen to match as closely as possible the geographic regions available in the craniometric data set. Unfortunately, there are no skin color data available for Polynesian populations. In addition, since there are few published reports of skin reflectance from East Asian populations, the Asian sample was extended to include samples from Central Asia (Tibet and Nepal) at an equivalent latitude.

F_{ST} values were estimated using both geographic regions (F_{RT}) and local populations (F_{LT}) as the units of analysis, using methods described elsewhere (Williams-Blangero and Blangero, 1989; Relethford and Blangero, 1990; Relethford et al., 1997) and my computer program RMET (available on request). F_{ST} estimation requires an estimate of average heritability. For craniometrics, I used an average heritability of 0.55, as derived in my previous work (Relethford, 1994). For skin reflectance, I used a heritability of 0.66 for the E.E.L. 609 filter, as reported by Williams-Blangero and Blangero (1992).

Estimates of F_{RT} and F_{LT} were corrected for sampling bias (Relethford et al., 1997) and used with Equation (6) to derive variance components. Standard errors for F_{RT} and F_{LT} were computed using the method described by Relethford et al. (1997), and used to derive standard errors for the variance components following conventional statistical methods:

$$\begin{aligned} se(AR) &= se(F_{RT}) \\ se(WR) &= se(F_{RT}) \\ se(AL) &= \sqrt{[se(F_{LT})]^2 + [se(F_{RT})]^2} \\ se(WL) &= se(F_{LT}) \end{aligned} \quad (7)$$

RESULTS

Estimates of regional (F_{RT}) and local (F_{LT}) differentiation for craniometric and skin color data are reported in Table 2. Two different sets of analyses were performed, the first using all available geographic regions (g = 6 for craniometrics, and g = 5 for skin color), and the second restricted to the three major geographic regions of Sub-Saharan Africa, Europe, and East Asia (Central/East Asia for skin color). All estimates of F_{RT} and F_{LT} are large relative to their standard errors, indicating significant differentiation at both the regional and local level of analysis. In all cases, F_{LT} is larger than F_{RT}, as expected, since differentiation among local populations on a global level includes both variation among

TABLE 2. Estimates of regional (F_{RT}) and local (F_{LT}) population differentiation (standard errors are in parentheses)

Data set	Analysis	F_{RT}	F_{LT}
Craniometrics	6 regions	0.1458 (0.0014)	0.2125 (0.0015)
	3-region subset[1]	0.1136 (0.0018)	0.1655 (0.0020)
Skin color	5 regions	0.8789 (0.0007)	0.9109 (0.0006)
	3-region subset[2]	0.8715 (0.0008)	0.9103 (0.0006)

[1] Analysis confined to Sub-Saharan Africa, Europe, and East Asia.
[2] Analysis confined to Sub-Saharan Africa, Europe, and Central/Eastern Asia.

TABLE 3. Variance components (standard errors in parentheses)

	Craniometrics, number of regions		Skin color, number of regions	
Source of variation	6	3	5	3
Among regions	0.1458 (0.0014)	0.1136 (0.0018)	0.8789 (0.0007)	0.8715 (0.0008)
Within regions	0.8542 (0.0014)	0.8864 (0.0018)	0.1211 (0.0007)	0.1285 (0.0008)
Among local populations	0.0667 (0.0021)	0.0519 (0.0027)	0.0320 (0.0009)	0.0388 (0.0010)
Within local populations	0.7875 (0.0015)	0.8345 (0.0020)	0.0891 (0.0006)	0.0897 (0.0006)

TABLE 4. Comparison of variance components from blood polymorphisms, DNA polymorphisms, craniometrics, and skin color

			Variance components (%)		
Data	Reference	Number of regions	Among regions (*AR*)	Among local populations within regions (AL)	Within local populations (WL)
Blood polymorphisms	Lewontin (1972)	7	6.3	8.3	85.4
Blood polymorphisms	Latter (1980)[1]	6	10.4	5.6	84.0
Blood polymorphisms	Ryman et al. (1983)	3	9.9	4.1	86.0
Microsatellite DNA	Barbujani et al. (1997)	5	10.0	5.5	84.5
RFLPs, 16 loci	Barbujani et al. (1997)	5	8.0	8.4	83.6
RFLPs, 79 loci	Barbujani et al. (1997)	4	11.7	3.9	84.5
Microsatellite DNA	Jorde et al. (2000)	3	10.4	1.7	87.9
RFLPs	Jorde et al. (2000)	3	13.2	1.3	85.5
Alu insertions	Jorde et al. (2000)	3	17.4	1.8	80.9
mtDNA (HVS1)	Jorde et al. (2000)	3	22.0	6.0	72.0
mtDNA (HVS2)	Jorde et al. (2000)	3	24.9	6.2	68.9
Y-chromosome[2]	Jorde et al. (2000)	3	7.8	5.1	87.1
Craniometrics	Present study	3	11.4	5.2	83.5
Craniometrics	Present study	6	14.6	6.7	78.8
Skin color	Present study	3	87.2	3.9	9.0
Skin color	Present study	5	87.9	3.2	8.9

[1] This study used three different methods that produced equivalent results; the figures reported use the method described by Lewontin (1972).
[2] Obtained when the highly divergent Finnish and Northern European populations were removed from analysis. See Jorde et al. (2000) for details.

regions as well as variation among local populations within regions. The most striking result in Table 2 is the very large estimates of F_{RT} and F_{LT} for skin color, indicating relatively low amounts of variation within local populations.

Table 3 reports the variance components for craniometric and skin color variation. Comparative values of AR, AL, and WL are reported in Table 4 for several studies of genetic markers (primarily red blood cell groups, and protein/enzyme polymorphisms) and DNA polymorphisms. The genetic estimates show considerably more variation within local populations (mean = 82%) than among local populations (mean = 5%) or among geographic regions (mean = 13%). These averages are somewhat inflated by the higher levels of among-region variation for mitochondrial DNA and *Alu* polymorphisms (for discussion, see Jorde et al., 2000). Excluding these estimates, the average variance components from genetic data are WL = 85%, AL = 5%, and AR = 10%.

The pattern shown by craniometric variation is strikingly similar to that seen in the genetic estimates; most variation is within local populations, followed by a much smaller proportion among regions, and still smaller proportions among local populations within regions. Roughly 79–83% of craniometric variation is among individuals within populations. The results for skin color are quite different from both genetic and craniometric results, and show the majority of variation (88%) occurring among geographic regions.

DISCUSSION

The apportionment of global genetic diversity estimated from craniometric data mirrors the pattern observed in several studies of genetic markers and DNA polymorphisms in showing that the majority of

diversity exists within local populations. Genetic data suggest that roughly 10% of total species diversity exists among major geographic regions, 5% among local populations within regions, and 85% within local populations. Craniometric variation follows the same basic pattern, with roughly 13% among regions, 6% among local populations within regions, and 81% within local populations. Comparison of the six-region and three-region analysis shows that these figures vary somewhat, depending on the specific groups analyzed.

The exact estimates will also vary depending on the specific estimate used for average heritability. Additional analyses using different values for heritability (not shown) reveal that a higher average heritability gives a lower proportion of variation among regions and a higher amount within local populations, whereas a lower average heritability gives the reverse. Although the choice of average heritability affects the exact estimates of the variance components, the overall pattern is the same even for a wide range of heritability estimates. For example, the three-region craniometric analysis provides estimates of roughly 80–90% variation within local populations for a range of average heritability from 0.4–1.0. In all cases, the results consistently agree with the evidence from genetic data that the vast majority of human variation exists within local populations.

The results from both genetic and craniometric analyses are consistent with neutral traits under an isolation by distance model. In the absence of selection and with some level of local endogamy, the variation among individuals within populations will exceed that among populations. The tendency for more variation to occur among regional populations than among local populations within regions is also a reflection of isolation by distance. According to isolation by distance models, genetic similarity among populations decreases exponentially as geographic distance increases. Thus, genetic differences between populations will be greater at the larger geographic distances separating regional groups than at the shorter geographic distances separating local populations.

The strong similarity between genetic and craniometric results suggests that global patterns of craniometric variation can be considered, on average, selectively neutral. I am not implying that craniometric variation is not affected by selection for *certain* traits, but instead that *multivariate* patterns of among-group variation produce genetic distances in agreement with neutral genetic markers. A similar argument has been made for genetic markers; although some individual genetic loci have been linked to natural selection, the average pattern over many loci tends to fit neutral expectations and provides valuable information about population history (Cavalli-Sforza et al., 1994). The same observation also applies to multivariate analyses of craniometric variation (e.g., Relethford and Harpending, 1994, 1995). Lahr (1996) suggested that selection can act on a number of different craniometric traits, such as those relating to overall size, such that similar patterns of among-group differentiation could result. While likely true to some extent, it is important to note that the data set used here includes a large number of variables from different parts of the face and skull, and is more likely to estimate underlying neutral patterns than a smaller number of interrelated variables. Further, a factor analysis of a subset of these data (Howells, 1973) shows a number of independent factors, suggesting that this data set provides information other than correlated size-related patterns.

The results based on global skin color variation are quite different, with the vast majority of total variation (88%) occurring among geographic regions, and much less occurring among (3%) or within (9%) local populations. This pattern is not unexpected, given the strong evidence of natural selection affecting global variation in skin color. Several studies have shown a strong correlation of skin color with latitude, with darker average skin color in populations living at or near the equator and increasingly lighter skin color with increasing distance from the equator (Roberts et al., 1976; Tasa et al., 1985; Relethford, 1997; Jablonski and Chaplin, 2000). The worldwide distribution of human skin color is correlated with the global distribution of ultraviolet radiation, suggesting past selection for dark skin near the equator and light skin at greater latitudes, north and south. Although debate continues over the exact selective agents (e.g., skin cancer, sunburn, vitamin D synthesis, or photolysis of folate), the implications are clear in terms of variance components. The greatest differences will occur across large differences in latitude (e.g., Europe and Sub-Saharan Africa), thus leading to the greatest proportion of total variation occurring among regions.

Skin color has often been used as a major characteristic for classifying human populations into different races. The race concept implies similarity within geographic regions, which does apply for skin color (although the geographic pattern of skin color variation is clinal, as opposed to a discrete pattern expected under the race concept). The apportionment of skin color variation is atypical, and does not match the pattern seen in DNA polymorphisms, genetic markers, or craniometric traits. As such, skin color cannot be used to make inferences about the geographic variation of other traits. Although a case can be made for genetic and craniometric data being a reflection of population structure and history of neutral traits (on average), the distribution for skin color has been affected differentially by natural selection, and tells little about global population history and relationships. It is ironic that one of the most visible human characteristics and one that has dominated in racial classifications is the least illuminating about underlying patterns of global hu-

man genetic diversity. The high level of among-region variation seen in skin color does not apply to genetic or craniometric traits, thus weakening the case for using skin color as a proxy for other biological traits.

LITERATURE CITED

Barbujani G, Magagni A, Minch E, Cavalli-Sforza LL. 1997. An apportionment of human DNA diversity. Proc Natl Acad Sci USA 94:4516–4519.

Brown RA, Armelagos GJ. 2001. Apportionment of racial diversity: a review. Evol Anthropol 10:34–40.

Cavalli-Sforza LL, Menozzi P, Piazza A. 1994. The history and geography of human genes. Princeton: Princeton University Press.

Falconer DS. 1981. Introduction to quantitative genetics. 2nd ed. London: Longman.

Howells WW. 1973. Cranial variation in man: a study by multivariate analysis of patterns of difference among recent human populations. Papers of the Peabody Museum no. 67. Cambridge, MA: Harvard University Press.

Howells WW. 1989. Skull shapes and the map: craniometric analyses in the dispersion of modern *Homo*. Papers of the Peabody Museum no. 79. Cambridge, MA: Harvard University Press.

Jablonski NG, Chaplin G. 2000. The evolution of human skin coloration. J Hum Evol 39:57–106.

Jorde LB, Watkins WS, Bamshad MJ, Dixon ME, Ricker CE, Seielstad MT, Batzer MA. 2000. The distribution of human genetic diversity: a comparison of mitochondrial, autosomal, and Y-chromosome data. Am J Hum Genet 66:979–988.

Lahr MM. 1996. The evolution of modern human diversity: a study of cranial variation. Cambridge, UK: Cambridge University Press.

Latter BDH. 1980. Genetic differences within and between populations of the major human subgroups. Am Nat 116:220–237.

Lewontin RC. 1972. The apportionment of human diversity. Evol Biol 6:381–398.

Nei M. 1987. Molecular evolutionary genetics. New York: Columbia University Press.

Nei M, Roychoudhury AK. 1982. Genetic relationship and evolution of human races. Evol Biol 14:1–59.

Relethford JH. 1994. Craniometric variation among modern human populations. Am J Phys Anthropol 95:53–62.

Relethford JH. 1997. Hemispheric difference in human skin color. Am J Phys Anthropol 104:449–457.

Relethford JH. 2000. Human skin color diversity is highest in Sub-Saharan African populations. Hum Biol 72:773–780.

Relethford JH, Blangero J. 1990. Detection of differential gene flow from patterns of quantitative variation. Hum Biol 62:5–25.

Relethford JH, Harpending HC. 1994. Craniometric variation, genetic theory, and modern human origins. Am J Phys Anthropol 95:249–270.

Relethford JH, Harpending HC. 1995. Ancient differences in population size can mimic a recent African origin of modern humans. Curr Anthropol 36:667–674.

Relethford JH, Crawford MH, Blangero J. 1997. Genetic drift and gene flow in post-famine Ireland. Hum Biol 69:443–465.

Roberts DF, Kahlon DPS. 1976. Environmental correlations of skin colour. Ann Hum Biol 3:11–22.

Rogers AR, Harpending HC. 1983. Population structure and quantitative characters. Genetics 105:985–1002.

Ryman N, Chakraborty R, Nei M. 1983. Differences in the relative distribution of human gene diversity between electrophoretic and red and white cell antigen loci. Hum Hered 33: 93–102.

Stringer CB, Andrews P. 1988. Genetic and fossil evidence for the origin of modern humans. Science 239:1263–1268.

Tasa GL, Murray CJ, Boughton JM. 1985. Reflectometer reports on human pigmentation. Curr Anthropol 26:511–512.

Williams-Blangero S, Blangero J. 1989. Anthropometric variation and the genetic structure of the Jirels of Nepal. Hum Biol 61:1–12.

Williams-Blangero S, Blangero J. 1992. Quantitative genetic analysis of skin reflectance: a multivariate approach. Hum Biol 64:35–49.