Human genetics

All humans belong to the same species, *Homo sapiens*; yet there are many populations, or races, that can be distinguished from each other in terms of morphological characters. The broad division into three major races, Caucasoid, Negroid, and Mongoloid, is generally accepted; many anthropologists add two more major races, Amerind (American Indians) and Australoid. The genetic differences between races are caused by the differences in gene frequencies at various loci. Therefore, by examining racial differences in gene frequencies, the genetic relationship and evolution of human races can be studied.

Gene frequencies are the relative frequencies of alleles at a locus in a population. For example, the ABO blood group locus has three major alleles, A, B, and O, and the frequencies of alleles A, B, and O in the English population are 25%, 6%, and 69%, respectively. These frequencies vary from population to population. In the early days of anthropological studies, the genetic relationship among different races was studied by examining the geographical distribution of gene frequencies at a few polymorphic loci such as the ABO and Rh blood group loci. It was later realized, however, that comparison of gene frequencies for one or two loci is not reliable, since each locus has a different distribution. Only when a large number of loci are examined does a genetic relationship become clear. This is partly because genetic variation between races is very small compared with the variation within races at the gene level. However, if a large number of loci are examined, even small differences can be detected with sufficient accuracy.

The genetic difference between a pair of populations is usually measured by a quantity called genetic distance, which is a function of gene frequencies. Once genetic distances are computed for a group of populations or races, their genetic relationships can be studied quantitatively. For this purpose, gene frequency data for loci that are detectable by electrophoresis or antigen-antibody reaction are usually used. In addition, some investigators have begun to use deoxyribonucleic acid (DNA) polymorphism data for this purpose.

Electrophoretic and antigenic loci. The results obtained from studies of electrophoretic and antigenic loci suggest that the Caucasoid and Mongoloid races may be genetically closer to each other than either is to the Negro race (see illus.). However, this is not conclusive because the number of loci used is not sufficiently large. The genetic distances between populations within each of the three major races are generally much smaller than those between the major races. Particularly, European populations (such as English, French, Italian, Basque, and Finnish) are very closely related. In general, there is a tendency for geographically close populations to show small genetic distance. This is apparently due to gene migration that has occurred in the past, or to recent splitting of the populations. Unlike the view generally held by anthropologists, the populations belonging to Amerind do not cluster with those belonging to Mongoloid. This is apparently due to genetic drift (random fixation of alleles) that occurred in the tribal Amerind populations. Amerind populations show large genetic distances even among each other. Similarly, the tribal populations belonging to Australoid show large genetic distances.

There are several factors that affect the genetic differentiation of human races, two of which are mutation and isolation. A well-defined race often has variant alleles that are unique to the race. For example, African Negroid people have many unique alleles such as $F^4$ at the Duffy locus and $PepC^5$ at the peptidase C locus. This suggests that the Negroid population has been isolated from other groups for a long time. Similar unique alleles have been found in several other populations, notably Australoid and South American Indian populations. Migration has an effect opposite to that of isolation. In the process of human evolution, gene migration seems to have occurred rather extensively in some groups of populations. For example, Europeans and Iranians are genetically quite close, though they live geographically far apart. Past migration can often be traced back by examining the distribution of rare alleles.

Language differences can be a barrier to interpopulational hybridization. Indeed, there is often a correlation between genetic distance and linguistic distance when a group of closely related populations are studied. However, the correlation between genetic distance and linguistic distance at the racial

level is quite small. This is apparently because language is culturally inherited and can change rapidly under certain circumstances.

The correlation between genetic distance and morphological difference is also generally quite weak. For example, the Negritos and aboriginal Malays in Southeast Asia and Pygmies and Bushmen in Africa have a number of common morphological features such as short stature, dark skin, and frizzy hair. Because of these similarities, some anthropologists believed that they originated from the same common stock. Genetic distance data, however, have shown that the African and Southeast Asian populations are genetically quite different and are more closely related to their respective neighboring populations. Clearly, the evolutionary change of morphological characters is quite different from that of electrophoretic and antigenic loci. Morphological characters seem to be subject to stronger natural selection than average genes.

**DNA polymorphism data.** Data on DNA polymorphism are still quite limited, but available data indicate that the extent of DNA polymorphism is much higher than variation at the protein level. It has been estimated that two randomly chosen genes at a locus have, on the average, about four nucleotide differences, even if only the coding region of DNA is considered. However, variation between races is again much smaller than variation within races. Mitochondrial DNA (mtDNA) is known to have no recombination, so that a phylogenetic tree of polymorphic mtDNAs can be constructed. A number of authors have constructed such a tree, and the results obtained indicate that the divergence times of some pairs of polymorphic mtDNAs are very old, that is, much older than the time of the origin of *Homo sapiens* (about 200,000 years ago) or the time of divergence of the three major races. It has also been shown that although there is some tendency for the mtDNAs sampled from the same race to cluster, most of them are genealogically mixed with those from other races. Therefore, mtDNA data are not sufficient for discerning the evolutionary relationships of different races. To clarify these relationships, information on DNA polymorphism for many independently inherited nuclear genes seems to be necessary.

For background information see Anthropology; Human Genetics; Population Genetics; Recombination (Genetics) in the McGraw-Hill Encyclopedia of Science and Technology.

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