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HUMAN GENETIC VARIATION: THE MECHANISMS AND RESULTS OF MICROEVOLUTION

ABSTRACT

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This paper presents an overview of the patterns of human genetic variation and the processes that generate them. There are three principal goals. The first is to introduce the new concepts, terms, and methods that have emerged to meet the needs of direct observations on DNA. The second is to look at the extent and pattern of variation in human DNA sequences, and how a record of our evolution is embedded within the patterns of variation. The final objective is to evaluate whether or not race concepts are consistent with new findings. A brief introduction to the coalescent model is given. The coalescent model is an integrative framework for understanding patterns of variation in terms of the processes that generate them. Two data sets with DNA observations on humans from different regions of the world are discussed. Both data sets indicate that human populations differ in the amount of variation that they harbor. At most genetic loci, African populations harbor some relatively common alleles that are absent in non-African populations; however, most of the alleles that are common in non-African populations are also common in African populations. Thus, the pattern of genetic variation is one of nested subsets, such that the variation in non-African populations is a subset of the variation found in African populations. The recent out of Africa model, which postulates an ancient founder event that occurred with the human occupation of new continents, is the best current explanation for this pattern of variation. It is concluded that the typical definitions of human races do not fit the patterns of variation in DNA sequences very well, but it is difficult to get a firm handle on race because race concepts are often used in vague and imprecise ways. In conclusion, the architecture of human genetic variation is explained by the evolutionary history of our species and is best understood in that context.