

The Genomic Distribution of Human Population Substructure: Basic and Applied Human Population Genomics

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Abstract:

Human genetic variation is increasingly being recognized as an important source of both statistical power and potential confounding in complex disease studies. There are several aspects of genetic variation that can clarify the potential for these two factors to adversely and/or beneficially affect study designs. The first is to understand the important axes across which there is genetic variation within the population to be studied. For many contemporary populations there is variability in individual ancestry levels brought about through the process of genetic admixture – the intermixture between previously isolated populations. Admixture can be used in a number of ways to study the genetics of complex disease. Second, it is important to understand that evolution has not affected all genomic regions to the same extent. The practical application of admixture gene mapping and phenotype investigation highlight the general utility of an understanding of the evolutionary histories of human populations with particular reference to the extent to which particular genomic regions have evolved.