

# Ancient Admixture in Human History

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**ABSTRACT** Population mixture is an important process in biology. We present a suite of methods for learning about population mixtures, implemented in a software package called ADMIXTOOLS, that support formal tests for whether mixture occurred and make it possible to infer proportions and dates of mixture. We also describe the development of a new single nucleotide polymorphism (SNP) array consisting of 629,433 sites with clearly documented ascertainment that was specifically designed for population genetic analyses and that we genotyped in 934 individuals from 53 diverse populations. To illustrate the methods, we give a number of examples that provide new insights about the history of human admixture. The most striking finding is a clear signal of admixture into northern Europe, with one ancestral population related to present-day Basques and Sardinians and the other related to present-day populations of northeast Asia and the Americas. This likely reflects a history of admixture between Neolithic migrants and the indigenous Mesolithic population of Europe, consistent with recent analyses of ancient bones from Sweden and the sequencing of the genome of the Tyrolean “Ice-man.”

**A**DMIXTURE between populations is a fundamental process that shapes genetic variation and disease risk. For example, African Americans and Latinos derive their genomes from mixtures of individuals who trace their ancestry to divergent populations. Study of the ancestral origin of the admixed individuals provides an opportunity to infer the history of the ancestral groups, some of whom may no longer be extant. The two main classes of methods in this field are local ancestry-based methods and global ancestry-based methods. Local ancestry-based methods such as LAMP (Sankararaman *et al.* 2008), HAPMIX (Price *et al.* 2009), and PCADMIX (Brisbin 2010) decompose ancestry at each locus in the genome and provide individual-level information about ancestry. While these methods provide valuable insights into the recent history of populations, they have reduced power to detect older events. The most commonly used methods for studying global ancestry are principal component analysis (PCA) (Patterson *et al.* 2006) and model-based clustering methods such as STRUCTURE (Pritchard *et al.* 2000) and

ADMIXTURE (Alexander *et al.* 2009). While these are powerful tools for detecting population substructure, they do not provide any formal tests for admixture (the patterns in data detected using these methods can be generated by multiple population histories). For instance, Novembre *et al.* (2008) showed that isolation-by-distance can generate PCA gradients that are similar to those that arise from long-distance historical migrations, making PCA results difficult to interpret from a historical perspective. STRUCTURE/ADMIXTURE results are also difficult to interpret historically, because these methods work either without explicitly fitting a historical model or by fitting a model that assumes that all the populations have radiated from a single ancestral group, which is unrealistic.

An alternative approach is to make explicit inferences about history by fitting phylogenetic tree-based models to genetic data. A limitation of this approach, however, is that many of these methods do not allow for the possibility of migrations between groups, whereas most human populations derive ancestry from multiple ancestral groups. Indeed there is only a handful of examples of human groups in which there is no evidence of genetic admixture today. In this article, we describe a suite of methods that formally test for a history of population mixture and allow researchers to build models of population relationships (including admixture) that fit genetic data. These methods are inspired by the ideas by Cavalli-Sforza and Edwards (1967), who fit phylogenetic

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# Ancient Admixture In Human History Genetics

**Ian Pickup**



## **Ancient Admixture In Human History Genetics:**

## Decoding **Ancient Admixture In Human History Genetics**: Revealing the Captivating Potential of Verbal Expression

In a period characterized by interconnectedness and an insatiable thirst for knowledge, the captivating potential of verbal expression has emerged as a formidable force. Its capability to evoke sentiments, stimulate introspection, and incite profound transformations is genuinely awe-inspiring. Within the pages of "**Ancient Admixture In Human History Genetics**," a mesmerizing literary creation penned by way of a celebrated wordsmith, readers embark on an enlightening odyssey, unraveling the intricate significance of language and its enduring impact on our lives. In this appraisal, we shall explore the book's central themes, evaluate its distinctive writing style, and gauge its pervasive influence on the hearts and minds of its readership.

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