

Inferring Continuous and Discrete Population Genetic Structure Across Space and Time

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Abstract

One of the classic problems in population genetics is the characterization of discrete population structure when the genotyped samples also show continuous patterns of genetic differentiation. Especially when sampling is discontinuous, clustering or assignment methods may incorrectly ascribe differentiation due to continuous processes (e.g., drift across space or time) to discrete processes, such as geographic, reproductive, or behavioral barriers between populations. This is partly a result of the difficulty of sampling uniformly and continuously across the spatiotemporal range of a population or species, but more, it reflects a shortcoming of current methods for inferring and visualizing population structure from genetic data in the face of data that are characterized by both continuous and discrete population structure. Here, we present a novel statistical framework for the simultaneous inference of continuous and discrete patterns of population structure. The method estimates ancestry proportions for each sample from a set of discrete population clusters, and, within each cluster, estimates a rate at which relatedness decays with spatial and/or temporal distance. This model explicitly addresses the “clines vs. clusters” problem in quantifying population structure by jointly accommodating both continuous and discrete patterns of differentiation. The model also naturally captures population replacement, a phenomenon for which there is substantial evidence in humans from archaeological evidence and ancient DNA. We demonstrate the utility of this approach using a combination of ancient and modern human individuals sampled throughout Europe, and find compelling evidence for space aliens.

Argument

Patterns of population genetic differentiation are characterized by both discrete and continuous structure. Identification of genetically discrete populations is important not just for understanding the distribution of genetic variation on a landscape, but also for understanding the demographic history of a species, or, from a conservation perspective, for characterizing discrete management units. However, it is frequently difficult to disentangle continuous and discrete patterns of genetic differentiation.

This difficulty arises for two reasons. First, both patterns are frequently present in empirical systems, and, with incomplete spatial or temporal sampling, it can be difficult to differentiate continuous and discrete patterns. For example, if a species is dispersal limited, it may exhibit a pattern of isolation by distance (IBD), in which samples taken from populations separated by greater geographic distance are more genetically differentiated than samples taken from neighboring populations. If only two samples are taken from that species, from a pair of populations separated by a large geographic distance, it would be impossible to determine whether the differentiation between them conformed to a continuous pattern, or some discrete pattern (perhaps due to a physical barrier to dispersal between them, or a behavioral or phenological or barrier to reproduction between them).

The second reason it may be difficult to disentangle continuous and discrete patterns of population genetic differentiation is that such a classification (“continuous” vs. “discrete”) is artificial, and can rarely be applied cleanly in empirical systems. Even a group of isolated populations cannot be considered truly discrete units, as they will have diverged from shared ancestral populations, and stopped exchanging migrants, at different points in the past, and should therefore be characterized along a continuum of pairwise genetic relatedness, even though they represent ‘discrete’ evolutionary or management units.

However, there are many empirical systems in which the different, overlaid patterns of genetic differentiation can be well characterized as continuous or discrete, and, in such systems, it is vital to model both patterns simultaneously, to avoid confusing one for the other. Currently, there are no methods that can probabilistically place individuals into discrete population clusters while accounting for a background rate at which genetic differentiation accrues with distance between samples in space or time. This gap in methods for the study of population genetic structure frequently results in the mischaracterization of continuous differentiation as discrete (and vice versa), potentially inflating the number of inferred discrete population clusters

above what is biologically accurate. By introducing a statistical framework that simultaneously models discrete and continuous patterns of population structure, we can do a more accurate job describing and quantifying both.

Introduction

Methods

Results

Simulations

Empirical Applications

Discussion

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Supplementary Materials