

Demographics of genetic admixture and expansion

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Motivation and Aim: We study the dynamics of genetic admixture and expansion, as well as language assimilation, through mathematical-demographic modelling. Our primary aim is to address the 'paradox' in population genetics, where autosomes and allosomes can provide markedly different, if not contradictory, pictures of past migrations. More broadly, we aim to develop a model setup that would allow the reconstruction of past demographic parameters that might have produced a given population genetic outcome.

Methods and Algorithms: We develop population reproduction models for each of the types of genetic markers studied: a single-sex heterogeneous population model for uniparental markers (allosomes) and two-sex heterogeneous population models for autosomes and for the linguistic assimilation model. To facilitate modeling the long-term population change without unnecessary complications due to age structural change, we use the reproductive value [1, 2] as our main indicator of contribution to the population genome. Our input variables are the initial population sizes, reproductive advantage of the initial elite and marriage selectivity parameters.

After developing the formalism for the proposed demographic models, we perform numerical simulations aimed at emulating two scenarios stylised after two empirical populations. The first empirical population is that of the Beaker Complex people of Britain [3], whose migration to Britain led, according to palaeogenetic data, to a rapid and almost complete turnover of the population genome. The second empirical population is that of the Karachay-Balkar people, two closely related Turkic-speaking North Caucasian autochthons who show a complex population genetic pattern: while their Y-DNA shows a contribution of about one third of steppe origin, their autosomal DNA is no different from that of their Caucasian neighbours and shows only a minor contribution attributable to East Turkic migrations [4–6].

Results: We develop formal relations for the processes of expansion of the allosomes and of admixture of autosomal DNA using, in the latter case, both selective and non-selective marriages scenarios. To examine consequences of social adaptation, we also develop models of linguistic assimilation similar to the models of genetic admixture. We demonstrate that the aforementioned paradox may find a purely demographic explanation, as single-sex and two-sex reproduction models exhibit markedly distinct dynamics. We illustrate that the three processes – allosomal expansion, autosomal admixture, and language assimilation – occur at significantly different modes, potentially explaining the varied outcomes of these processes upon the completion of ethnogenetic transitions.

The genetic expansion process, which describes the evolution of non-recombinant allosomes, proceeds rapidly by historical standards. Demographically, this expansion process can be effectively modeled as a single-sex heterogeneous population.

Even more rapid is the process of autosomal admixture, when intermarriages, even if limited, are permitted. This admixture, essentially modeled as a two-sex reproduction process, leads to the swift convergence of both “invaders” and “autochthones” in terms of autosomes. Consequently, the autosomal genetic makeup stabilizes quickly and becomes independent of the allosome expansion. This decoupling explains why population genetic makeups often exhibit significant divergence between allosomes and autosomes. The rapidity of genetic admixture suggests that the initial reproductive advantage (or disadvantage) of immigrants may rapidly diminish through social assimilation processes, which we modeled using linguistic assimilation formalism. By designating one language as “dominant” (either superstrate or substrate) and assuming that every child with a parent fluent in that language adopts it, our model captures the essence of social assimilation. While real-life assimilation may be slowed by cultural norms or accelerated by schooling and social interaction, our findings demonstrate that the assimilation process can occur swiftly, even faster than genetic admixture. This phenomenon of social assimilation explains why initial reproductive differentials can quickly dissipate, halting genetic expansion and leading to complex population genetic make-ups at the end of ethnogenesis.

Our numerical illustrations demonstrate that the formal models proposed offer valuable insights into reconstructing migrations and mechanisms of ethno-genesis in the past. The first illustration supports the hypothesis in the literature that the Beaker Complex people likely arrived in Britain during a period of sparse population. Simulations based on the genetic makeup of the Karachay-Balkars lend credibility to the scenario of elite dominance in their formation. Our results suggest a small (up to about five per cent) initial migration of powerful elites who had a strong reproductive advantage but freely intermarried with the local population.

Building upon formal results and numerical simulations, we propose several qualitative indicators of modes of social transformation in historical populations. A large disparity between the proportions of invading genome in allosomes and autosomes serves as an indicator of quick admixture, suggesting minimal obstacles to interethnic marriages. Conversely, similar proportions of invading genome in allosomes and autosomes indicate lack of mixed marriages. A limited eventual proportion of invading allosomes suggests quick social assimilation and ethno-genesis, highlighting the rapid integration of immigrant populations into the existing social fabric.

Conclusion: Our research offers valuable insights into the intricate interplay between demography, genetics, and social organization, providing implications for historical scenarios and enhancing our understanding of the long-term consequences of migration and social cohesion.

The fluidity of linguistic transitions, even in comparison to population genetic transitions, underscores the need for caution when employing linguistic arguments to study ethnicity over extended historical periods.

Another practical implication of our results is that while the genetic composition of the population after the expansion process is complete may show a substantial contribution from the newcomers, especially in the allosomes, their initial share of the genome may be much smaller. This is all the more true for the population share, as the per capita reproductive value of immigrants would typically be higher than that of the autochthonous population due to the younger age composition. This suggests that it may be difficult to trace the original migration events using direct palaeogenetic evidence, especially if the first generations of invaders buried their deceased in their former place of origin.

Our findings highlight the need for deeper, more nuanced population genetic analyses that do not focus solely on autosomal or allosomal genes. Analysing different aspects of a population's genetic make-up together will provide more robust conclusions and more meaningful insights into the population's past.

Last but not least, our models may be useful for better understanding the population genetic consequences of modern migrations in large cities or developed countries, where migration has increased to levels that affect population reproduction [7].

Our models can be extended in several ways to improve our understanding of past migrations: by modelling both Y and MtDNA simultaneously, by considering stochastic and diffusion effects, by assuming multiple or even continuous migrations.

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