Available online at www.sciencedirect.com

ScienceDirect

Current Opinion in Genetics & Development

Worldwide genetic and cultural change in human evolution

Nicole Creanza[1,2] and Marcus W Feldman[1]

Both genetic variation and certain culturally transmitted phenotypes show geographic signatures of human demographic history. As a result of the human cultural predisposition to migrate to new areas, humans have adapted to a large number of different environments. Migration to new environments alters genetic selection pressures, and comparative genetic studies have pinpointed numerous likely targets of this selection. However, humans also exhibit many cultural adaptations to new environments, such as practices related to clothing, shelter, and food. Human culture interacts with genes and the environment in complex ways, and studying genes and culture together can deepen our understanding of human evolution.

Addresses
[1] Department of Biology Stanford University, Gilbert Hall, 371 Serra Mall, Stanford, CA 94305, United States
[2] Department of Biological Sciences, Vanderbilt University, 465 21st Ave. South, Nashville, TN 37212, United States

Corresponding author: Creanza, Nicole (nicole.creanza@vanderbilt.edu)

Current Opinion in Genetics & Development 2016, **41**:85–92

This review comes from a themed issue on **Genetics of human origin**

Edited by **Joshua M Akey** and **Anna Di Rienzo**

For a complete overview see the Issue and the Editorial

Available online 16th September 2016

http://dx.doi.org/10.1016/j.gde.2016.08.006

Introduction

The study of worldwide genetic variation has made great strides in the 25 years since researchers first convened to plan the Human Genome Diversity Panel (HGDP) [1]. The initial analyses of HGDP data showed that the vast majority of genetic variation occurs within human populations; however, the small fraction of between-population genetic variation could be used to characterize clusters of individuals, which generally correspond to geographic regions and can often be further segmented into population-level groups [2]. The data produced as a result of this initiative, combined with the HapMap and 1000 Genomes initiatives and additional samples from modern and ancient populations, continues to shed light on important aspects of human evolution, including demographic history, migration patterns, admixture between groups, selection pressures, and mutation rates [3••,4•,5–10].

Meanwhile, it has become increasingly clear that human culture interacts with genetic variation in complex ways. Culture can evolve through similar processes to genetic evolution: cultural variants can have differential survival and reproduction, but there are notable differences between cultural transmission, mutation, and inheritance and their genetic analogues [11–13]. Cultural transmission does not obey the precise rules that Mendelian inheritance imposes on single genes, and it may occur between unrelated individuals. Culturally transmitted traits, such as norms and preferences, can change within the course of a human generation, and cultural inheritance may occur over many generations, between groups rather than individuals, and depend on the environmental or social context in which an individual lives. Further, genes and culture often interact: several researchers have suggested that genetic changes, for example those that affect brain architecture, can promote large-scale changes in human culture [14,15], but cultural changes can also alter the selective advantage of genetic mutations, fostering their spread [16–18]. In one classic example, the spread of dairy farming and animal domestication in multiple geographic regions led to a corresponding regional increase in the frequency of genetic variants associated with lactase persistence, allowing more individuals to benefit from drinking milk into adulthood [19,20]. This interaction between genetic and cultural evolution has been studied under several research umbrellas, including gene–culture coevolution, dual inheritance theory, and cultural niche construction [19,21,22]. Here, we review the literature on human genetic and cultural variations, the interactions between them, and the importance of considering both genes and culture in studies of human evolutionary history.

Patterns of worldwide genetic variability and the influence of cultural practices

Geographic patterns of human demographic history have left detectable signatures on the human genome. For example, the human migration out of Africa likely occurred by repeated founder events, in which a small group of people broke away from a larger population to establish a new settlement [23]. Since each subsequent founder event constitutes a sample of the genotypes of the larger group, the serial founder effect model predicts a decrease in genetic diversity with geographic distance from the putative human origin in Africa [24]. Patterns of human

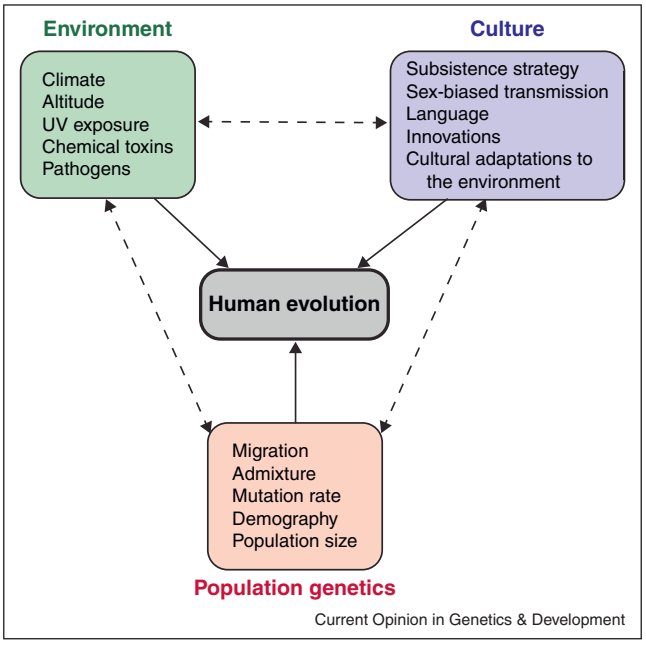
genetic variation have also shed light on the extent of admixture between different populations [25]. This admixture can be a result of relatively recent events in human history, such as colonialism or the advent of technology that facilitates long-distance transportation [4•,8,25,26]. However, recent studies have illustrated that ancient admixture events, such as between modern humans and Neanderthals or Denisovans, are also detectable in the modern human genome [27••,28••].

As researchers accumulate genetic data from more human populations and develop more sophisticated computational techniques, the effects of various forces in population genetics — for example, recent population growth [29], population separation [30•], range expansion [31], neutral genetic variation [32,33], and mutational load [34•] — can be understood in much greater detail. However, signals of population genetic and demographic processes in the human genome are complicated by cultural factors. For example, runs of homozygosity (ROH) are stretches of the genome where heterozygous nucleotides are absent or extremely rare, indicating that an individual's two chromosomes share a recent ancestor, with the length of each run dependent on the number of generations since the common ancestor [35]. ROH can provide evidence for population bottlenecks and ancestral relationships between populations, but it is important to note that the length of these runs can be dramatically influenced by cultural practices, particularly those surrounding marriages between relatives [35–37]. Indeed, homozygosity-based measurements can be used to estimate inbreeding more accurately than can be achieved with family pedigrees [38], particularly in cases where parental relatedness is elevated for many generations [39]. This inbreeding can, in turn, be negatively associated with phenotypes that are relevant to fitness and health, such as height, educational attainment [40], and hypertension [41].

Consanguinity and the cultural practices surrounding it provide one example of a culturally transmitted behavior that leaves an identifiable signature on the human genome. Other aspects of human culture, such as religion [42,43] and sex-specific demographic features [44] including sex-biased migration and sex-specific definition of cultural belonging, can also shape a population's trajectory of genetic evolution. By separately tracing the evolution of maternally transmitted mitochondrial sequences and paternally transmitted Y chromosomes, researchers can test the genetic effects of cultural practices such as matrilineality and patrilineality [45], as well as other sex-biased patterns of human demography that are culturally determined (Figure 1).

For example, the deep phylogenetic history of mitochondrial DNA sequences suggests that human populations were matrilineally structured before the out-of-Africa expansion [46]. A further study of Eurasian and African populations found a discrepancy between the Y chromosome and mitochondrial DNA in the signal of expansion events, implying that male gene flow might have been restricted in some ancestral lineages [47]. Marital practices in which a man relocates to his wife's village upon marriage have left a genetic signature of reduced effective population size and genetic diversity for females in Timor [48]. In contrast, patrilineal societies exhibit male-biased transmission of reproductive success, likely culturally transmitted, which leads to reduced genetic diversity [49]. The sex-specific cultural practices surrounding age of reproduction can also leave a mark on genetic variation, with faster matrilineal genetic evolution in Iceland attributed to a shorter generation interval in women [50]. In the specific example of the Hindu caste system, the cultural tradition of hypergamy, in which women are permitted to marry into a higher social caste in some circumstances but men are not socially mobile, has led to female-specific gene flow and, in some cases, genetic stratification of the populations [51]. Thus, societal systems and cultural norms can have an affect on genetic evolution; however specific cultural events can also leave a mark on the genome. For example, known migration events or significant cultural innovations in human history may correspond to dramatic expansions of the male human lineage, detectable on the Y chromosome [52••]. Although some of the effects of culture on

Figure 1



Genetic, environmental, and cultural factors are capable of influencing one another (dashed arrows), and all three have an impact on human evolution (solid arrows).