

# Hunter–gatherer genomics: evolutionary insights and ethical considerations

Richard J Bankoff<sup>1,2</sup> and George H Perry<sup>1,3</sup>



Hunting and gathering societies currently comprise only a small proportion of all human populations. However, the geographic and environmental diversity of modern hunter–gatherer groups, their inherent dependence on ecological resources, and their connection to patterns of behavior and subsistence that represent the vast majority of human history provide opportunities for scientific research to deliver major insights into the evolutionary history of our species. We review recent evolutionary genomic studies of hunter–gatherers, focusing especially on those that identify and functionally characterize phenotypic adaptations to local environments. We also call attention to specific ethical issues that scientists conducting hunter–gatherer genomics research ought to consider, including potential social and economic tensions between traditionally mobile hunter–gatherers and the land ownership-based nation-states by which they are governed, and the implications of genomic-based evidence of long-term evolutionary associations with particular habitats.

## Addresses

<sup>1</sup> Department of Anthropology, Pennsylvania State University, University Park, PA 16802, USA

<sup>2</sup> Intercollege Program in Bioethics, Pennsylvania State University, University Park, PA 16802, USA

<sup>3</sup> Department of Biology, Pennsylvania State University, University Park, PA 16802, USA

Corresponding authors: Bankoff, Richard J ([rjb398@psu.edu](mailto:rjb398@psu.edu)) and Perry, George H ([ghp3@psu.edu](mailto:ghp3@psu.edu))

Current Opinion in Genetics & Development 2016, 41:1–7

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

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

<http://dx.doi.org/10.1016/j.gde.2016.06.015>

0959-437/© 2016 Elsevier Ltd. All rights reserved.

## Introduction

Before the advent of agriculture in Southwest Asia ~12,000 years ago, every human society practiced locally adapted forms of hunting and gathering [1]. Using both modern and ancient genomic techniques, research on the global spread of agriculture and agriculturalist populations from multiple independent centers of domestication has uncovered complex, regionally specific histories of admixture and replacement between agriculturalist and

hunter–gatherer societies [2–5]. While many hunter–gatherers adopted new domesticates and became agriculturalists [6], and others appear to have been entirely replaced by agricultural migrants from elsewhere [7], approximately 230 hunter–gatherer societies have maintained this lifestyle through the present day, or at least until the very recent past [8]. Extant hunter–gatherer population sizes are now orders of magnitude smaller than their agriculturalist neighbors, but some maintain high levels of genetic diversity compared to agriculturalist populations, reflecting larger ancestral populations [9], and many hunter–gatherer groups have inhabited their distinct local environments for far longer than their agriculturalist neighbors [10,11]. In still other cases, hunter–gatherers have inhabited environments not conducive to intensive agriculture, such as the high arctic. Thus, hunter–gatherers offer an incredible opportunity to study human adaptations to local environments [12].

While the evidence of environment-specific adaptation in hunter–gatherers from genomic studies is a major focus of this review, we also consider and account for the ethical implications of this research. Marginalized by the spread of agricultural and industrialized societies, modern hunter–gatherers have often been confined to liminal environments worldwide, from the Arctic tundra to the rainforests of equatorial Africa and the Australian desert. Because these societies are inherently reliant on a mobile lifestyle to procure their subsistence, many of the world's remaining hunter–gatherers are threatened by the imposition of political boundaries and property rights that severely restrict their ability to access traditional resources. The richness of the hunter–gatherer genetic heritage combined with the exclusion of these societies from many of the formal institutions of the nation-states that control their traditional homelands suggests that they should be considered vulnerable populations by bioethicists and genomic researchers [13–15]. Here we review the recent literature on ethical considerations of genomics research in hunter–gatherer populations. In particular, we focus on issues of informed consent, community engagement, and the culturally and politically sensitive presentation of results from evolutionary genomic studies.

## Methods to detect candidate signatures of selection in hunter–gatherer genomes

Recent studies of hunter–gatherer populations have employed generalized scans for positive natural selection to investigate how the varied and often extreme conditions

of their local environments may have shaped phenotypic adaptation. Potential signatures of positive selection include unusual shifts in patterns of haplotype variation (i.e., extensive linkage disequilibrium) surrounding a candidate locus, an unusual allele frequency spectrum at a candidate locus, or an unusual degree of between-population allele frequency differentiation relative to the genome-wide distribution for these values [16]. With the increasing feasibility of generating large population genomic datasets, these analytic tools have been used to identify candidate signatures of natural selection in multiple hunter–gatherer populations, as described below.

### How do hunter–gatherer adaptations to local environments manifest in the genome?

Among genes located within or nearby genomic loci highlighted by scans for positive selection in worldwide hunter–gatherer populations, several biological pathways recur. First, loci implicated in immune function and parasite suppression appear to have been under strong positive selection (i.e., with signatures of classical selective sweeps on the basis of patterns of haplotype variation, and/or high levels of allele frequency differentiation compared to other populations) in multiple hunter–gatherer populations [17–21]. This finding is not unexpected, given the overrepresentation of immune loci in selection scans in any human population [22,23]. However, compared to agricultural populations, hunter–gatherers experience distinct pathological and parasitological burdens related to significant differences in population density, sedentism, and habitat use. For instance, hunter–gatherer populations in India possess particular variants of the interleukin-4 gene associated with anti-parasitological activity at much higher frequency than their agriculturalist neighbors, possibly as a result of living with higher burdens of helminthic endoparasites [24]. In the Aka, a rainforest hunter–gatherer population from the Congo basin, it has been hypothesized that positive selection has driven to high frequency a genetic variant that confers a reduced efficiency of the CYP2A6 nicotine-processing enzyme, because this phenotype would lengthen the residence time of nicotine in the bloodstream and lower total parasite loads [25].

Other sets of gene ontologies repeatedly overrepresented in scans for positive selection in worldwide hunter–gatherer populations are those involved in metabolism, body mass, and body shape. For example, Siberian and Greenlandic hunter–gatherers appear to have convergently evolved separate metabolic mechanisms for subsisting on a highly carnivorous diet in an extremely cold climate. First, in northeastern Siberia, Chukchi and Koryak populations exhibit one of the strongest selective sweep signatures yet discovered in humans, centered on a functional variant of the long-chain fatty-acid oxidation gene *CPT1A* [26<sup>•</sup>]. This variant seems to have provided a selective advantage among people with a diet high in

animal fats, despite also being associated with increased infant mortality risk due to hypoketotic hypoglycemia [27]. Second, in Greenland, a recent population genomic study of a Greenland Inuit population identified a very strong signature of positive selection on the basis of extreme inter-population allele frequency differentiation near three fatty acid desaturase (*FADS*) genes [28<sup>•</sup>]. Three SNPs in particular appear to play an outsized role in regulating height and body mass, perhaps related to the adaptive utility of a high body-mass to surface area ratio in the extreme cold of the Arctic, as suggested by Bergmann and Allen’s rules [29,30]. Interestingly, the alleles affected by positive selection in the Inuit are also present at low frequencies in non-hunter–gatherer European and East Asian populations, but their associations with height and body mass have been missed in large genome-wide association studies (GWAS) due to their rarity outside of the Greenland population [28<sup>•</sup>].

Arctic populations are not the only hunter–gatherers who display signatures of body mass evolution. One of the most striking examples of environment-specific human body shape variation is the ‘pygmy phenotype,’ or small adult body size (e.g., average adult male stature <155 cm), a characteristic common to hunter–gatherer populations inhabiting tropical rainforests in Africa [20] and Asia [19,31,32]. A number of scholars have hypothesized that this phenotype represents an ecological adaptation to the challenging rainforest habitat, potentially providing an evolutionary advantage by reducing metabolic demands in a calorically impoverished environment, easing demands of thermoregulation, improving mobility in the dense undergrowth and for climbing trees, speeding life histories to increase reproductive potential for individual with a high disease burden, or some combination thereof [4,33,34]. Scans for strong positive selection on stature-related loci in rainforest hunter–gatherer populations have suggested that this phenotype may result from independent adaptations on multiple pathways in different populations, including the growth hormone IGF1 pathway [19,20], genes involved in pituitary development and insulin-adiponectin signaling [5], and bone homeostasis and remodeling [17]. In a different approach, Perry *et al.* [35] performed a GWAS to identify 16 genetic loci associated with the pygmy phenotype in BaTwa rainforest hunter–gatherers from Uganda, and then identified a signature of polygenic adaptation (a distributed signature of positive selection) at these loci.

### What can these patterns of adaptation tell us about human evolution and variation?

The diverse lifestyles and ecologies of modern hunter–gatherer societies, while changed by interactions with agriculturalists over the past 12,000 years, are still substantially more representative of those experienced by humans over the vast majority of our evolutionary history

Download English Version:

<https://daneshyari.com/en/article/5892895>

Download Persian Version:

<https://daneshyari.com/article/5892895>

[Daneshyari.com](https://daneshyari.com)