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A paleogenetic perspective on the early population history of the high altitude Andes

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ABSTRACT

The peopling of the high altitude Andes marks an important episode in South American population history, eventually leading to the formation of the most complex societies of the late pre-Columbian period, namely Wari, Tiwanaku, and Inca. Little is known about how population dynamic processes and genetic adaptation to physical stressors like hypoxia shaped the genetic diversity of the Andean highlanders over the ~12,000 years of human presence in high altitude.

Here we review the genetic population history of the Central Andes and how ancient DNA research helps provide a more nuanced understanding of the early peopling of the high altitude Andes. We further discuss the potential of paleogenetic analyses to address questions of Andean archaeology and human adaptability to stress factors acting at high altitude. The reviewed genetic data support broader genetic continuity in the Central Andes for at least 8000 years, with intra-Andean population dynamics processes subsequently shaping the regional genetic structure, and connects the populations of the Pacific Littoral with the early populations of the high altitude Andes.

1. Introduction

The high altitude Central Andes were the heartland of some of the most complex pre-Columbian societies. The establishment of late pre-Columbian states like the Inca, the Tiwanaku, and the Wari can be attributed to the ability of ancestral Andeans to cope physically and economically with the specific environmental conditions they faced >2500 m above sea level (m.a.s.l.). It is widely accepted that humans began to live seasonally at high altitudes shortly after the archaeological record indicates human presence along the Central Andean coastline c.13,000 to 14,000 years ago (Dillehay et al., 2012; Sandweiss, 1998). These initial signs of human activity in the high altitude Andes have been interpreted as transhumant expeditions of foragers from the pacific littoral expanding their access to resources (Aldenfelder, 2008; Sandweiss, 1998). However, Andean archaeologists have long debated when humans began to live permanently at high altitude. As discussed in other contributions in this volume, humans living at high altitude face an environment characterized by hypoxic, i.e. low concentration of atmospheric oxygen, and cold conditions that negatively impact the abundance of resources available to sustain human populations (Aldenfelder, 2008). Both extremes act as stressors on the human body affecting biological processes, e.g. reproductive growth, basal metabolic rates, morbidity, and mortality (Beall, 2014; Moore et al., 2004). Arguments against early permanent settlement of the high Andes include the need for a long period of adaptation to the extreme environmental conditions (cf. Aldenderfer, 2006). These assumptions are challenged by recent findings of potentially year-round Terminal Pleistocene high altitude sites like the Cuncaicha rock shelter in the Pucuncho Basin, in the Southern Peruvian highlands, discussed by Rademaker et al. in this volume (Rademaker et al., 2014, 2013). The early high altitude sites suggest that permanent residence as high as 4000 m.a.s.l. was established c. 12,000 to 10,000 years ago; recent work, however, challenges a year-long occupation of the site (Capriles et al., 2016). Nevertheless, a time period of roughly 2000 years from the onset of human presence along the coast to permanent habitation of the high Andes is often considered too brief to establish the adaptive phenotypes and underlying genotypes observed in modern Andean highlanders, e.g. biological adaptation to hypobaric hypoxia (Beall,

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2014; Bigham et al., 2010; Moore, 2001; Zhou et al., 2013).

In recent years, genetic studies of present-day Amerindian populations have provided clues about both the genetic adaptation to high altitude stressors in Andean populations (e.g. Bigham et al., 2010; Eichstaedt et al., 2014; Zhou et al., 2013) and, more generally, the genetic population history of South America and the Central Andes (e.g. Lewis, 2009a,b; Reich et al., 2012; Scliar et al., 2012; Yang et al., 2010). Still, a question central to understanding how humans permanently colonized the Central Andes highlands remains unanswered: did exposure to hypoxia drive adaptation via natural selection, or did an existing phenotype—characterized by reduced susceptibility to hypoxic stress—enable the human settlement of the Andes? The estimation of genetic diversity and structure necessary to address this question is currently lacking, partly due to an unresolved origin and demographic history of Andean highlanders. While there is a growing consensus that the initial occupants of the western Andes arrived from the Pacific littoral (cf. Lewis et al., 2007a,b; Rothhammer and Dillehay, 2009), others propose an initial trans-altiplanic entry from the north (Lynch, 1990), or later westward expansions of Amazonian populations into the Andes (Moraga et al., 2001; Rothhammer et al., 2001).

The potential to address the questions of high-altitude adaptation solely by studying living populations is limited because of the unknown impact of ancient demographic processes on the genetic structure and diversity of Andean populations. The period of European contact and subsequent colonialism led to massive population decline and population admixture in the Americas (cf. Livi-Bacci, 2006). Additionally, the arrival of Europeans altered adaptive landscapes via the introduction of new economic systems, technologies, and also pathogens (cf. Crosby, 2004; Livi-Bacci, 2006). The analysis of ancient DNA (aDNA) from archaeological human remains provides a unique opportunity to directly explore the genetic diversity of pre-contact Andean populations to address the highlanders' population history and the evolutionary processes of adaptation to high altitude. Because understanding adaptation to high altitude requires a deeper understanding of population genetics in general, this chapter will review what is currently understood about the population history and demography of Central Andean populations, while the mechanisms of high altitude adaptation are reviewed in Lorna Moore's contribution in the same volume.

2. Ancient DNA studies in the Central Andes

One of the most recent and remarkable achievements in aDNA research is the ability to generate complete mitochondrial and/or nuclear genomes from prehistoric and historic human populations (Fu et al., 2016; Haak et al., 2015; Llamas et al., 2016; Mathieson et al., 2015; Raghavan et al., 2014). By sequencing genomic data from individuals spanning thousands of years, evolutionary geneticists can infer the timing of macro and micro-evolutionary processes involved in human adaptations to environmental and dietary changes (e.g. Fehren-Schmitz and Georges, 2016; Mathieson et al., 2015; Sverrisdóttir et al., 2014; Wilde et al., 2014).

In South American aDNA research, mitochondrial haplogroup assignments from about 700 individuals and mitochondrial hypervariable region (HVR) haplotype data from about 400 individuals have been reported over the past 20 years (cf. reviews by Raff et al., 2011; Fehren-Schmitz et al., 2012). Assigning a mitochondrial sequence to one of the four Native American mitochondrial haplogroups — A, B, C, or D — requires, in some cases, as few as a single polymorphism, reflecting only a broad unit of genetic diversity. The HVR region, on the other hand, is ~400 bp stretch of continuous sequence information from the D-Loop segment of the mitochondrial genome that is known to accumulate diversity in a way that allows a finer resolution within a haplogroup – referred to as a haplotype. The classic HVR sequencing approach nonetheless only covers a small proportion of the ~16,500 bp mitochondrial genome, thus mtDNA genetic diversity estimated from this region alone might be underestimated. Furthermore, mitochondrial DNA is only inherited from the mother, it can also only represent female population dynamics. The number of aDNA studies specific to the Central Andes has increased in recent years and ancient genomic data are increasingly available. Within only the last 12 months, more than hundred complete mitochondrial genomes from pre-Columbian Central Andean individuals were published (Fehren-Schmitz et al., 2015; Gómez-Carballa et al., 2015; Raghavan et al., 2105; Llamas et al., 2016; Valverde et al., 2016), illustrating the speed of progress and the direction of future aDNA research in the Central Andes. Furthermore, paleogenetic investigations in the highlands have been limited until recently to populations younger than ~1500 years (Raff et al., 2011). The latest studies, e.g. Early Holocene human remains from Cueva Lauricocha, now extend the time depth of available aDNA data in the high altitude Andes to ~8500 BP (Fehren-Schmitz et al., 2015) and in Southern Chile to ~9000BP (Reyes et al., 2012). Similarly, the geographic sampling coverage has increased massively in recent years, with aDNA data now representing the entire coastal and highland Central Andean region (Fig. 1). Still, the aDNA data available for South America are largely biased towards mitochondrial DNA, an artifact of the development of paleogenetic methods. Mitochondrial DNA are non-recombining, uniparentally-inherited genetic markers. While they are easier to isolate and analyze due to their abundance and short molecular length, they shed light only on maternally-driven population processes. Genomic-level data, inherited by both parents, are naturally more informative but much more difficult to acquire, partly due to poor DNA preservation in the region, and partly due to the sheer size and complexity of the human genome, roughly 180,000 times as large as mitochondrial genomes. Only two publications to date report low coverage genome-wide data from pre-Columbian South American individuals (Carpenter et al., 2013; Raghavan et al., 2015); one other study simply reported preliminary results (unpublished conference communications). The only complete human genomes reported thus far derive from North American individuals (Rasmussen et al., 2015, 2014).

Nevertheless, the small-scale studies that target relatively few nuclear and mitochondrial markers continue to offer powerful information to physical anthropologists and archaeologists. Using limited numbers of mitochondrial, autosomal and Y-chromosomal markers, researchers have been able to address questions of ancestry, kinship, and social change in the Andes (Baca et al., 2012; Carnese et al., 2010; Fehren-Schmitz et al., 2015), as well as the evolutionary consequences of exposure to environmental stress factors (de Rubira et al., 2015; Fehren-Schmitz and Georges, 2016; Georges et al., 2012).

While still far from rivaling the magnitude of paleogenomic data reconstructed for other continental regions like Western Eurasia, aDNA research is steadily on the rise for the Central Andes. The following chapters will continue to discuss how aDNA informs our understanding of Andean population history and human adaptation.

3. The peopling of the Central Andes from a genetic perspective

The investigation of the genetic ancestry of Andean populations is strongly correlated with the debate surrounding which routes the first humans took during the initial dispersal into and throughout South America. Two prominent scenarios have

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