

NEWS AND VIEWS

PERSPECTIVE

The flickering genes of the last mammoths

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Woolly mammoths, *Mammuthus primigenius*, are arguably the most iconic of the extinct Pleistocene megafauna, and an abundance of large permafrost-embedded bone and ivory material (Fig. 1) means they were also among the first to yield credible DNA sequences (Hagelberg *et al.* 1994; Hoss *et al.* 1994). Despite mammoth remains being numerous throughout northern Eurasia and North America, both the earliest and most recent fossils are found in northeast Siberia, with the last known population being confined to Wrangel Island in the Arctic Ocean from around 10,000 years ago until their extinction around 4,000 years ago. The extent to which these Holocene mammoths were descended from the Pleistocene populations of Wrangel Island and the demographic nature of their terminal decline have, until now, remained something of a mystery. In this issue of *Molecular Ecology*, Nyström *et al.* (2012) report the first use of autosomal variation to track the decline of the last mammoths and, in doing so, take a significant step towards resolving these questions. The authors genotyped four microsatellite loci in 59 Pleistocene and Holocene mammoths from Wrangel Island and Chukotka in mainland northeastern Siberia and showed that while the Pleistocene-to-Holocene transition is associated with a significant reduction in genetic diversity, subsequent levels of variation remain constant until extinction. Such a pattern is somewhat surprising as it indicates that while the last mammoths were confined to only a few Arctic islands, their final extinction on Wrangel Island was not a gradual process resulting from loss of genetic diversity/inbreeding. Instead, it seems they maintained a viable effective population size of around 500 until near their presumably rapid extinction. While the ultimate agent of mammoth extinction remains unknown, the work of Nyström *et al.* (2012), suggests that we should be looking for something sudden, like a rapid change in climate/ecology or perhaps the arrival of humans.

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Since the first publication of mammoth mitochondrial DNA (mtDNA) sequences (Hagelberg *et al.* 1994; Hoss *et al.* 1994), a variety of ‘mammoth DNA’ studies have been published. These studies fall into four main types based on the nature of the genetic data produced and the kinds of questions addressed. Early studies were confined to mtDNA—until recently, the target of choice for ancient DNA studies—and focused on phylogenetic questions: principally, the position of mammoths in relation to the living elephantids. However, it soon became clear that because ancestral elephantid populations were likely to be large but speciation times were likely to be close, stochasticity in the genealogical process meant that even perfectly resolved mtDNA trees may not reflect the true species tree (Thomas *et al.* 2000). This led to the second type of study, where the phylogenetic position remained the focus but inferences were based on genomic data from multiple loci (Rohland *et al.* 2010), a task made considerably easier by the development of the next-generation sequencing (NGS) technologies that led to the publishing of a draft mammoth genome (Miller *et al.* 2008). The third type of study, also based on genomic data and frequently employing NGS, was focused on functional variation. Such studies showed that mammoths were polymorphic in coat colour, with both dark- and light-haired individuals co-occurring (Rompler *et al.* 2006; although also see Workman *et al.* 2011), and that they carried variants of haemoglobin that were better adapted to offloading oxygen to respiring cells at low temperatures (Campbell *et al.* 2010).

Meanwhile, mtDNA continued to be the locus of choice for those concerned with population-level inference of the demographic history of the species. This is because ancient mtDNA data are usually easier and cheaper to produce than ancient genomic data for the large samples required for most demographic inference and because a number of new statistical inference methodologies were developed for mtDNA that accommodate the ‘noisy’ nature of the genealogical and mutation processes (e.g. Drummond & Rambaut 2007). Such studies have revealed signatures of range expansion and Pleistocene population stability, but also major mtDNA lineage loss (Barnes *et al.* 2007), and population flux between Eurasia and North America (Debruyne *et al.* 2008). But until now, population-level demographic inferences using multiple genomic loci—particularly concerning mammoth extinction—are notably absent in the literature. This is somewhat surprising because while phylogenetic and functional studies are undoubtedly of wide interest, the impact of rapid climate change on species diversity and extinction is surely a more pressing



Fig. 1 Sergey Vartanyan during field collection, holding a woolly mammoth tusk found along a river embankment in northeast Siberia (photographer: Diana Solovyeva).

subject of scientific enquiry. Furthermore, while patterns of mtDNA variation contain important information on a species' demographic history, the quantity of single locus information is limited, and for more detailed inferences, it is necessary to turn to multiple nuclear loci (Balloux 2010).

Two phases in the demographic decline of mammoths deserve particular attention: the end-Pleistocene loss of continental populations, and the mid-Holocene extinction of populations on St Paul Island in the Bering Sea (around 6400 years ago) and Wrangel Island in the Arctic Ocean (around 4,000 years ago). Nyström *et al.* (2012) focused on the Wrangel Island population but also included a continental sample. Their samples cover the last 60,000 years and include the last known mammoth found to date. They found a dramatic reduction in microsatellite diversity at the Pleistocene–Holocene transition—a period in which mammoth remains are absent. Using a novel coalescent simulation approach, they inferred a large, stable and continuous northeast Siberian population up to the Pleistocene–Holocene transition—consistent with previous findings (Barnes *et al.* 2007)—followed by a more than tenfold reduction in effective population size as the mammoths' range contracted and they became isolated in the newly formed (through Holocene sea-level rise) Wrangel Island.

But perhaps more remarkably, there is no subsequent signature of population decline following the isolation of Wrangel Island mammoths, leading up to their extinction around 4,000 years ago. Indeed, using patterns of microsatellite variation, Nyström *et al.* (2012) infer that the effective population size remained at around 500 for this period.

Interestingly, this number is consistent with estimates of the Wrangel Island Holocene carrying capacity (149–819 mammoths), based on ecological modelling (Nyström *et al.* 2010), and is the proposed minimum effective population size required for an endangered species to retain evolutionary potential (Franklin & Frankham 1998). What caused the final demise of the Wrangel Island mammoths remains an open question. Human impact is a possibility, as is rapid climate/ecological change. But thanks to Nyström *et al.* (2012) it now seems that a gradual Holocene decline through inbreeding and a loss of diversity is considerably less likely.

Microsatellites are difficult to type even in the best-preserved ancient material but provide an excellent proxy for overall genetic diversity. The number of loci examined by Nyström *et al.* (2012) was relatively small, and richer nuclear data certainly exist for individual mammoths. But data quantity is a poor proxy for study quality, and genomic data should only rarely be considered an end in itself. In ecological and evolutionary studies, it is the questions and population-level inferences that are important, and when studying an extinct species, surely the important questions concern the extinction event itself. In these respects, this study marks only the beginning of our understanding of the process of mammoth extinction, but it is an important and indeed the first step in the right direction. More extensive data will follow, as will improvements in statistical inference methodologies, but ultimately there is no substitute for good questions and the right samples.

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M.G.T. is interested in evolutionary and population history inference using molecular and other data sources.

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