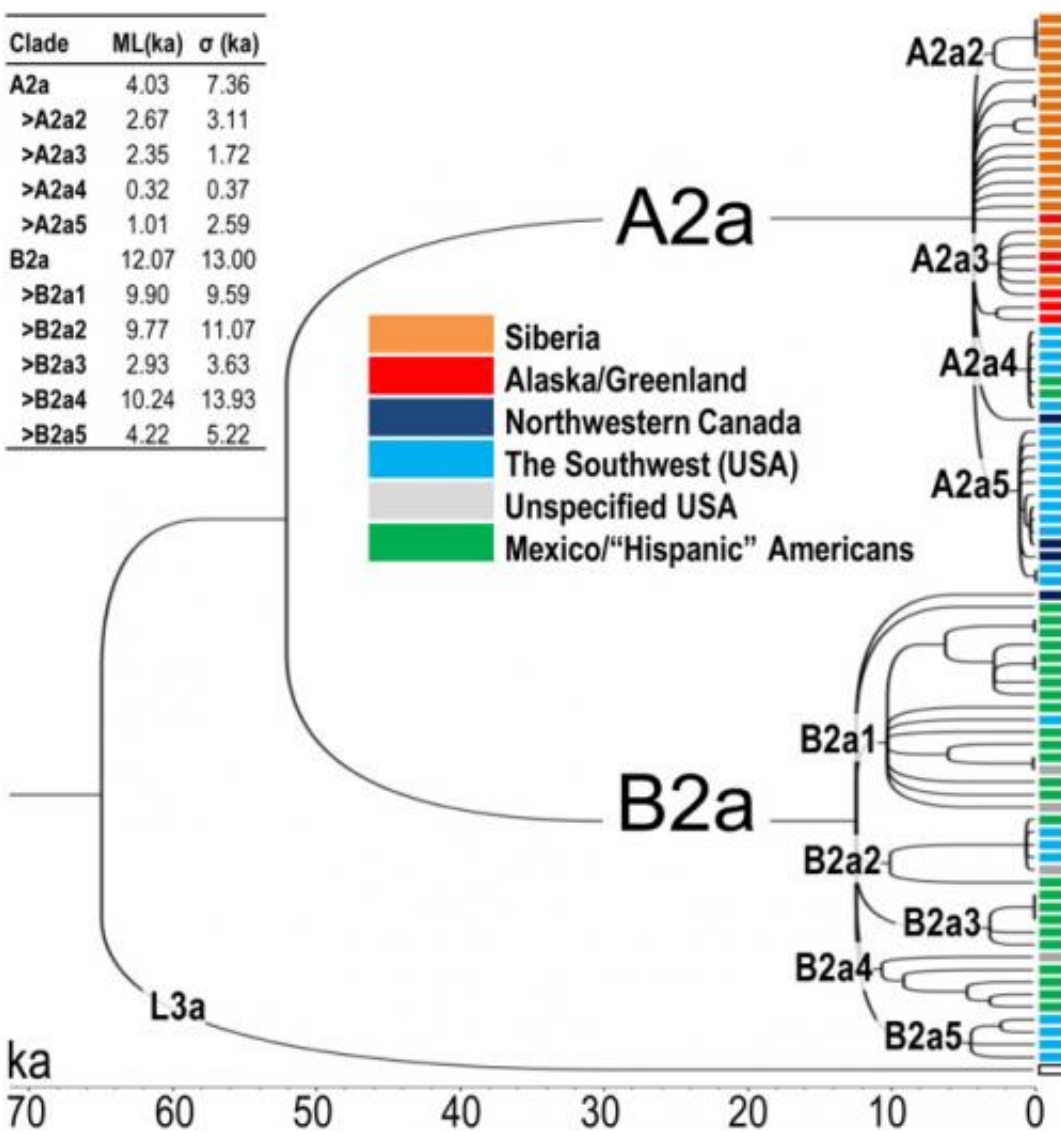


Getting here from there: Mitochondrial genome clarifies North American migration models

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Schematic phylogeny of complete mtDNA sequences belonging to haplogroups

A2a and B2a. A maximum-likelihood (ML) time scale is shown. (Inset) A list of exact age values for each clade. Credit: Copyright © PNAS, doi:10.1073/pnas.0905753107

(Phys.org) —It is generally agreed that the ancestors of modern Native Americans were Asian peoples who migrated to North America from Siberia and Beringia – a region proximate to the Bering Strait, Bering Sea and Chukchi Sea – over the so-called Bering Land Bridge, which was exposed, and therefore connected Asia with North America, at various times during the Pleistocene ice ages. At the same time, the question of whether this occurred through one or several streams of migration has long been a topic of considerable debate, with analyses based primarily on mitochondrial DNA (mtDNA) – which is directly inherited from in a maternal lineage – tending to coalesce around a model that hypothesized three distinct migratory streams (known as *tripartite migration*), which was originally proposed by combining anthropometric, genetic, and linguistic data. Recently, however, researchers at Università di Perugia and Università di Pavia, Italy (and a range of other institutions) evaluating these migratory models used mitochondrial genomes, or *mitogenomes*, to show that although the primary genetic signature was contributed by the first arrival, it was later modified not only by multiple additional migratory streams, but by local population dynamics as well. The scientists thus concluded that a standard three-wave model was too simplistic to account for the mitogenomic diversity revealed by their analysis.

Researcher Alessandro Achilli discussed the paper that he, Prof. Antonio Torroni and their co-authors published in the *Proceedings of the National Academy of Sciences (PNAS)*. "Over the previous decade there's been a renewed interest in studying the origin of [human populations](#), particularly [indigenous groups](#) of the New World," Achilli tells *Phys.org*.

"However," he notes, "there remains a level of disagreement among scientists regarding the number of migratory events and entry routes that gave rise to Native American groups. The general consensus is that modern Native Americans trace their ancestry to a limited number of original founders whose gene pool ultimately derived from Asians that peopled northeast Siberia and parts of Beringia prior to the Last Glacial Maximum, or LGM. (The LGM is a period in the Earth's climate history when ice sheets were at their maximum extension between 26,500 and 19,000–20,000 years ago.) The ancestral Beringian populations probably retreated into refugia (areas in which a population of organisms can survive through a period of unfavorable conditions) during the Ice Age, Achilli continues, where their genetic variation was reshaped not only because of drift, but also due to admixture with population groups newly arrived from regions located west of Beringia. "Therefore," Achilli explains, "pre-LGM mitochondrial haplotypes" – mtDNA variants inherited together from a female parent – "of Asian ancestry were differently preserved, modified, and lost in Beringian enclaves – and the main traces within the New World could be found within the closest regions, such as North America."

Regarding their molecular and phylogeographic mitogenome analyses, Achilli says that the team analyzed entire mitochondrial genomes belonging to two relatively rare Native lineages, known as A2a and B2a, which are characterized by peculiar geographic distributions. "A2a mtDNAs have been mostly identified only in Siberia, in Alaska and the surrounding regions, and in Natives from the American Southwest," he explains, "while B2a has also been observed exclusively in North America, just to the south and more widespread than A2a." A recent study¹, he adds, proposed that B2a mtDNAs mark a population expansion within the American Southwest that was due to the spread of groups of Uto-Aztecan farmers who introduced maize agriculture into this area from Mesoamerica about 4 kya (kiloannum, or thousand years, ago). "However, our analyses provide additional information that

suggests a different scenario, in which the onset of B2a should be predated to 11-13 kya in the late Pleistocene, and so is thus likely to be too ancient to be associated with the farming/language dispersal hypothesis."

Achilli also describes the key factors the scientists used to address these challenges. "We brought the analyses of two rare Native lineages, A2a and B2a, to the maximum level of molecular resolution – that of the entire mitogenomes – and analyzed its molecular variation by applying sophisticated statistical and phylogenetic tools," Achilli details. "Once that all Native American founder maternal lineages are identified and the entire mitochondrial genomes analyzed, it becomes possible to define both the entry time and the most likely entry route for each lineage." This allowed them to definitively resolve the issue of the number of distinct migrations that gave rise to modern Native American groups – and in particular to assess how many more recent migrations played a role in the formation of Native groups living in North America.

In addition to the first wave from Beringia about 15-18 kya, which followed the Pacific coastal route and rapidly reached South America, the researchers believe that more or less at the same time there was an inland migratory event, also from Beringia, which entered North America not along the coast but through the ice-free corridor in western Canada. "In contrast to the coastal founders," Achilli emphasizes, "this group of founders remained confined to northern North America, and contributed genetically to only the modern Amerindian groups living in that area, such as Algonquin and Na-Dene."

Much later, he adds – about 5 kya – there was another event: the spread of the carriers of haplogroup A2a from Alaska, undertaking both a westward migration to Asia and an eastward expansion into the circumpolar regions of Canada. "The carriers of A2a heavily contributed to the formation of the Paleo-Eskimos, but also mixed with the

descendants of the previous two migrations that contributed to the formation of modern Na-Dene. Even more recently," Achilli continues, "there were other events such as the spread of the Neo-Eskimo, or Thule, groups that probably brought the D3, and perhaps A2b1, mtDNAs from Alaska to South Greenland only one millennium ago. Thus, our analyses of mitogenomes from North America reveal at least four migratory events."

The next step in the team's research, Achilli notes, will be the analysis of the entire nuclear genome, starting from a more detailed analysis of the paternally inherited Y-chromosome. This will be possible due to new sequencing technologies that could be also applied to the DNA extracted from ancient bones.

In addition, there are other areas of research that might benefit from their study. "Our studies have always multidisciplinary features, since archeological evidence, as well as linguistic and climatic data, are extremely useful. Thus, the same research areas will also benefit from our findings," Achilli concludes, "but probably this particular paper will largely influence linguistics by reviving and perhaps going beyond the old tripartite linguistic subdivision of Native Americans into Eskimo-Aleuts, Na-Dene and Amerinds."

More information: Reconciling migration models to the Americas with the variation of North American native mitogenomes, *PNAS* August 27, 2013 vol. 110 no. 35 14308-14313, [doi:10.1073/pnas.1306290110](https://doi.org/10.1073/pnas.1306290110)

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¹Evaluating the Farming/Language Dispersal Hypothesis with genetic variation exhibited by populations in the Southwest and Mesoamerica, *PNAS* April 13, 2010 vol. 107 no. 15 6759-6764, [doi:10.1073/pnas.0905753107](https://doi.org/10.1073/pnas.0905753107)

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