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Human migration and population analysis

Before Christopher Columbus's famous voyage in 1492 and the beginning of modern transoceanic journeys, humans had established themselves on all habitable continents. This initial settlement of the continents by *Homo sapiens* originated approximately 200,000 years ago in Africa, and humans further dispersed into Eurasia, Oceania, and the Americas in the last 100,000 years. Those populations that still live in the same continent that their ancestors inhabited prior to 1492 are today referred to as indigenous or aboriginal populations. Thus, Europeans (for example, Basques and Germans) are indigenous or aboriginal populations of Europe, Africans (for example, San and Pygmies) are indigenous populations of Africa, and Native Americans are indigenous populations of the Americas. However, there are scant historical records of the origins of most of these populations, and, in any case, these records do not go back before 5000 years ago. Therefore, the prehistory of indigenous populations can only be deduced and reconstructed from vestiges investigated by different scientific disciplines.

To reconstruct prehistoric events, scientists have dealt mostly with material traces left by ancient cultures (archeology), fossil bones (physical anthropology), or cultural traits like language (linguistics). However, in the last few decades, there has been an increasing use of the genetic variability found in the DNA of living and dead individuals, which preserves an inherited record of ancestral populations. More recently, technological achievements that allow DNA recovery and the complete sequencing of genomes from ancient bones and tissues are providing unique information about the genetic variation of past populations. Indeed, the complete genomes of several Neandertals have been sequenced, revealing that around 2% of their DNA is present today among indigenous populations outside sub-Saharan Africa. In addition, a new hominin lineage related to Neandertals and modern humans, namely, the Denisovans, has been identified by sequencing a finger bone found in the Denisova cave in the Altai Mountains in Russia. Even though no known skulls or skeletons have been found to deduce their likely appearance, traces of 1–5% of Denisovan genes are also found among the indigenous populations of Oceania and Southeast Asia. More surprisingly, genome sequence comparisons have suggested that a third lineage of an ancient hominin, of whom almost nothing is known, may also have passed genes to current populations.

Population analysis. Some questions have traditionally been framed in terms of populations: Where and when did ancestral populations live? What was the route of migration of the ancestral populations that first occupied each continent? When did they arrive in their current homeland? Population-based questions of this kind assume that a population is a discrete entity that can be followed through time. If so, an individual's DNA will tend to be more similar to that of other individuals from the same popula-

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63 tion than to that of individuals from different pop-
64 ulations. The level of genetic similarity (or dissimi-
65 larity) between populations can be arranged along a
66 scale of time. Therefore, the more different that two
67 populations are, the older is the common ancestor
68 that they share. However, populations are not iso-
69 lated entities; there is recurrent migration of indi-
70 viduals between them, and some can be a product
71 of the blending of populations that were previously
72 isolated. Consequently, it may not be easy or even
73 meaningful to draw conclusions about populations
74 over long periods of time.

75 **Lineage and genomic analysis.** The molecular anal-
76 ysis of DNA has allowed alternative approaches to
77 the reconstruction of human history to be devel-
78 oped. Most of the genome is located on autosomes,
79 the non-sex chromosome pairs that are numbered
80 and named 1 to 22, which are always inherited by
81 the offspring from the mother and the father in
82 equal contributions. However, two special segments
83 of DNA show uniparental inheritance; that is, they
84 come exclusively from the father or the mother.
85 These segments are the Y chromosome, which is
86 transmitted only from fathers to sons, and the mito-
87 chondrial DNA (mtDNA), which is passed only from
88 mothers to children. They provide examples of DNA
89 lineages that can be followed over long periods of
90 time. The histories of different lineages may not be
91 identical because of chance or because of distinct
92 male or female contributions to a present-day pop-
93 ulation. Habits such as polygyny (many wives for a
94 man) and the assimilation of females from other pop-
95 ulations (for example, after a war) would be exam-
96 ples of factors leading to distinct histories of males
97 and females found in the same population. Small re-
98 gions of autosomes that are free of recombination
99 (a DNA exchange mechanism that generates new
100 combinations of genes derived from paternal and
101 maternal chromosomes) may also provide lineages
102 that can be followed through time. Genetic results
103 from the pregenomic period have led to the building
104 of a consensus scenario for the indigenous coloniza-
105 tion of the continents, which is now being extended
106 and complemented with DNA variation analysis from
107 complete genomes (**Fig. 1**).

108 The current level of genome technology allows
109 the generation of massive amounts of data for all
110 chromosomes from thousands of individuals, and
111 new analyses are being devised to investigate the
112 past based on the comparison of model scenarios of
113 population evolution. In these analyses, simulations
114 are used to model populations that are experienc-
115 ing different events (for example, migrations, de-
116 mographic expansions, or bottlenecks) at different
117 times. The population-genetic parameters obtained
118 for each simulated model are compared to the pa-
119 rameters estimated from the real genomic data, and
120 the most likely scenario is thus identified. As an ex-
121 ample, the impact of recent genetic studies on the
122 understanding of the origins of Native Americans
123 will be analyzed in detail.
124

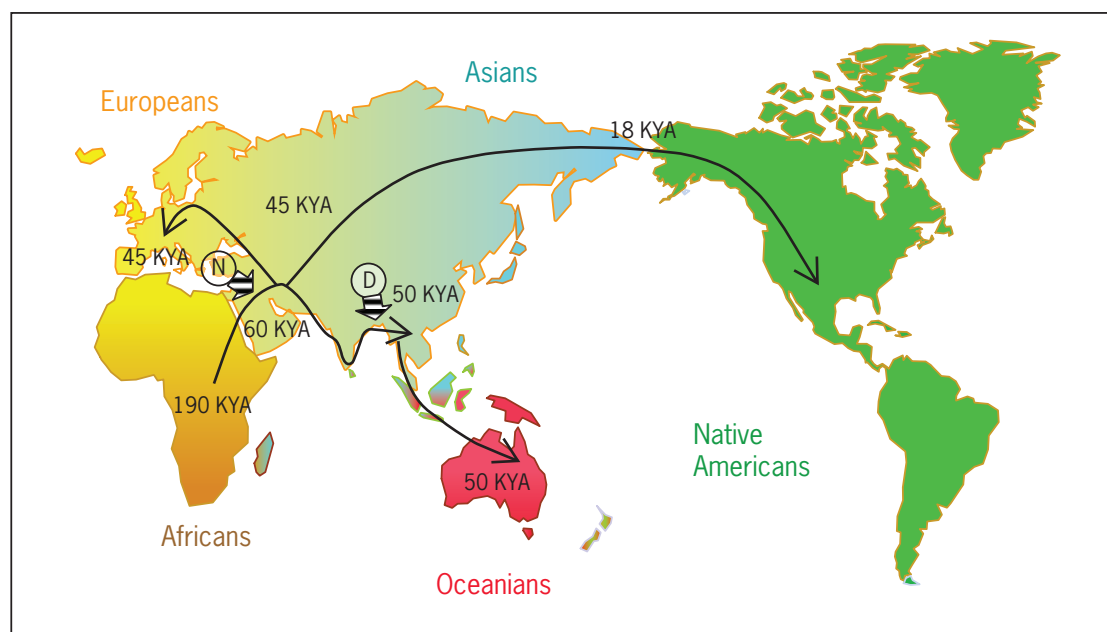


Fig. 1. Settlement of the indigenous world. This map represents a general view of the continental settlements by indigenous populations during human prehistory (black arrows). The anatomically modern humans, also called *Homo sapiens*, present an African origin, derived from a local population of *Homo erectus* at least 190,000 years ago (190 KYA). The out-of-Africa migration started 60 KYA, leading to the settlement of the major continental landmasses. Recent ancient genome evidence indicates a small contribution of DNA derived from Neandertals (N) and Denisovans (D) to populations outside Africa, which is probably the result of rare interbreedings between individuals of those species and modern human ancestors.

DNA history of the first journey to the New World.

When Europeans arrived in the Americas at the end of the fifteenth century, they found a group of people who were not previously known and had not been mentioned in the Bible. A Spanish Jesuit named José de Acosta was the first to suggest, in 1590, that the people now known as Native Americans were descendants of ancestors who had come from Asia a long time earlier. This idea is now generally accepted by scientists. In its modern form, it proposes that the ancestors of Native Americans came through Beringia, a land bridge between Alaska and Siberia that was located where the Bering Strait is now found; the bridge was formed when the sea level fell during glacial periods between 26,000 and 12,000 years ago (Fig. 2). The most likely pathway for the initial movement southward into the Americas was along the Pacific coast, around 16,000 years ago. Those individuals could have used small boats, and they fed on marine algae and animals along the ice-free coastline of North America.

A multidisciplinary hypothesis proposed in the 1980s claimed that the first Native Americans arrived from Siberia as recently as 12,000 years ago, and were followed by only two different groups at 9000 and 4000 years ago; this idea has now largely been refined by subsequent studies of archeology and genetics. The current scenario describes an initial migration from Northeast Siberia into Beringia (and North America) between 20,000 and 16,000 years ago, followed by one or several minor migration events between 14,000 and 4000 years ago, most likely by people using boats.

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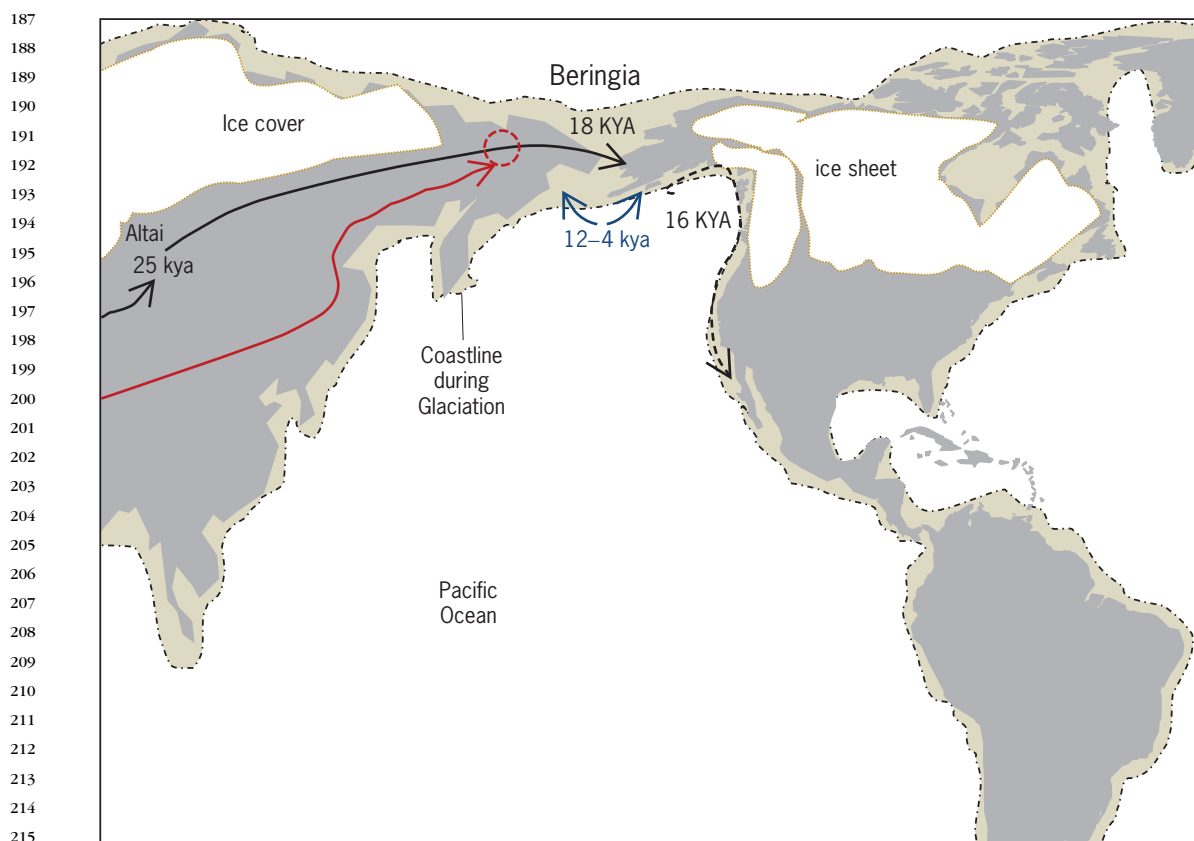


Fig. 2. The first peopling of the Americas. A current view of the indigenous settlement of the Americas based on genetic evidence indicates an occupation of Beringia around 18,000 years ago (18 KYA) by a Siberian-derived population (*black arrow*). According to recent data from ancient genomes, the gene flow between this Siberian-derived population and East Asians (*red arrow and circle*) generated a Beringian population. Other population-genetic studies, as well as morphology studies, indicate that recurrent gene flow from Siberia between 12 and 4 KYA brought new genes and the Sinodont morphology (*blue arrows*). The ancestors of Native Americans could have migrated southward around 16 KYA along the Pacific coastline by boat (*dotted black arrow*).

A current debate concerning the pre-Columbian peopling of the Americas involves the dynamics of the populations crossing Beringia. Studies involving uniparental markers (the Y chromosome and mtDNA) and skull morphology indicate that a major population coming from Central-South Siberia occupied Beringia around 18,000 years ago. This is illustrated by the existence of major Y chromosome and mtDNA lineages occurring frequently in all indigenous groups in the Americas. Still, there are some minor Y chromosome and mtDNA lineages found among current Native Americans, and there is also a morphological transition seen in East Asia and the Americas in ancient skulls dated between 12,000 and 6000 years ago that led from a generalized (un-derived) shape to a Sinodont (Mongoloid) shape. To explain these genetic and anatomical findings, it has been suggested that additional gene flow occurred in the last 12,000 years (Holocene), bringing some minor uniparental lineages and the typical Sinodont skull morphology to the Americas. However, very recent genomic data obtained from ancient bones from Central Siberia and North America have suggested that most DNA variation in the Americas is derived from a population that migrated from Northeast Asia,

249 resulting in a blend of populations from Central and
250 East Asia at around 18,000 years ago.

251 Even though studies using ancient genomes and
252 large population surveys involving uniparental mark-
253 ers and morphology appear to disagree in some ar-
254 eas, they present some concordant conclusions on
255 the timing of settlement and the Siberian origin of
256 Native Americans. Indeed, both genetic investiga-
257 tions indicate that the source population for Na-
258 tive Americans was located in Central Siberia (in the
259 vicinity of the Altai Mountains and Lake Baikal), and
260 that Native Americans share ancestors with western
261 Europeans through an older common ancestor in
262 Eurasia (or the Middle East).

263 **Conclusions.** The genetic evidence from uni-
264 parental and genomewide data suggests that all Na-
265 tive Americans are descended mostly from Siberi-
266 ans who arrived in the Americas toward the end
267 of the last glaciation in the Pleistocene. It reveals
268 an ancient migration of humans who came out of
269 Africa approximately 60,000 years ago and passed
270 through northern Eurasia (~40,000 years ago),
271 Siberia (~25,000 years ago), and Beringia (~18,000
272 years ago), from where they could migrate south-
273 ward to the Americas along the Pacific coastline
274 about 16,000 years ago (Fig. 2). *See also:* ANTHRO-
275 POLOGY; ARCHEOLOGY; DEOXYRIBONUCLEIC ACID
276 (DNA); EARLY MODERN HUMANS; FOSSIL HUMANS; HU-
277 MAN BIOLOGICAL VARIATION; MIGRATORY BEHAVIOR;
278 MOLECULAR ANTHROPOLOGY; NEANDERTALS; PALE-
279 OINDIAN; PHYSICAL ANTHROPOLOGY; POPULATION
280 DISPERSAL; POPULATION DISPERSION; POPULATION
281 GENETICS. Fabrício R. Santos; Chris Tyler-Smith

282 **Keywords:** DNA variability; human evolution; hu-
283 man migration; origin of indigenous populations;
284 settlement of Americas

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311 **URLs**

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313 <http://www.becominghuman.org/>

314 National Geographic: The Genographic Project

315 <http://www.genographic.com>

316 Smithsonian National Museum of Natural History:

317 Human Evolution

318 <http://humanorigins.si.edu>

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