The Peopling of the Americas: New Insights from Genetic Studies
Prof. Theodore G. Schurr

Outline of talk

- Anthropological background
  - Archeology, osteology, linguistics
- Genetics background
  - Markers and inheritance
- Molecular genetics data
  - mtDNA evidence
  - Y chromosome evidence
- Summary of data
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Pleistocene overkill hypothesis

Megafauna extinction models

- Human hunting
  - Human over-hunting directly caused the extinction, or perhaps eliminated a "keystone species" (usually the mammoths or mastodons), which, in turn, led to environmental collapse and a more general extinction
- Environmental causes related to climate change
  - Environmental changes might have caused extinction by eliminating food sources, disrupting birth schedules, or exposing animals to climatic conditions to which they were not adapted
- Hyperdisease
  - Humans brought with them one or more disease-causing agents that jumped from human or animal carriers (for example, their dogs) to the highly susceptible large native fauna
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Some Clovis-age archaeological sites, roughly 11,500-10,800 radiocarbon years ago (13,350-12,895 calendar years):
- Meadowcroft (PA) 16,800 cal BP
- Cactus Hill (VA) 16,000 cal BP
- Topper (SC) 16,000 cal BP

Some current candidates for pre-Clovis occupation sites:
Sources: Thomas R. Hester, E. James Dixon, Kenneth B. Tankersley

Cactus Hill
- Excavators: McAvoys
- Pre-Clovis dates from this site fall in the range of 15,070-16,940 cal ybp

Topper site
- Excavator: Alan Goodyear
- Pre-Clovis levels date to 16,000 cal ybp
- Claims new evidence for human occupation of 50,000 ybp (?)
Monte Verde
~14,850 cal BP

Monte Verde tools

Monte Verde

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Some Clovis-age archaeological sites; roughly 11,500-10,800 radiocarbon years ago (13,350-12,895 calendar years)

Some current candidates for pre-Clovis occupation sites

Monte Verde ~14,850 cal BP

Retreat of the glaciers
14,000 radiocarbon years ago (16,800 cal BP)
13,000 years ago (15,350 cal BP)
12,000 years ago (14,000 cal BP)
11,000 years ago (13,020 cal BP)
10,000 years ago (11,350 cal BP)
9,000 years ago (10,185 cal BP)

Dated animal remains and human occupations

No precursors to Clovis lithic culture in Siberia
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Paleoindian fluted point distribution


New coastal routes into the new world

Was Clovis first?
Possibly two different founding populations colonized the Americas, originating at different times and in different places in Asia.
Native American languages

- ~2,000 languages (extant + extinct)
- But how many language (foundling) stocks were brought to Americas?
  - Greenberg = 3 stocks: Amerind, Na-Dene, and Eskimo-Aleut
  - Others = 11-12 stocks within so-called Amerind, as well as Na-Dene and Eskaleut

Tripartite migration model

Possibly two migrations contributed to Amerind stock

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Summary of non-genetic data

- Archeological data beginning to reveal presence of pre-Clovis cultures in the Americas
- Craniometric data suggest expansions of two biologically distinct populations into Americas from Asia (early and later), but debated
- Linguistic data question unity of Amerind language stock, hence, single migration of ancestral Amerindians; also contested
- Geological and climatological data question availability of interior corridor route until 11,000-12,000 ybp, after emergence of Clovis culture, and accessibility of NW coast route

Inheritance patterns of different genetic systems

Control region
Gene map of the human mtDNA

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Phylogenetic analysis of mtDNA RFLP haplotypes and CR sequences

- Haplotype
- CR or HVR sequences
- Haplogroup, or lineage
- Outgroup

Human mtDNA haplogroup network

(Kivisild, et al., 1999)

Model of human mtDNA expansions across the world

Expansion times (years ago):
- East Asia: 50,000 - 60,000
- West Asia: 50,000 - 60,000
- Europe: 15,000 - 20,000
- Americas: 20,000 - 30,000
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Founding mtDNA lineages in Native Americans

Distribution of mtDNA haplogroups in the Americas
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mtDNA haplogroup frequencies in Siberia

mtDNA haplogroup ages in Siberia and the Americas

Pairwise mismatch distributions and population expansions

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Possible explanations for the apparent absence of 9RA in putative Asian source populations

9RA could have (i) arisen in the founding American population, (ii) been sampled from a putative Asian source population, in which it was subsequently lost by genetic drift, and (iii) been segregating in an Asian source population at the time of migration, but that source population has not been identified because it either went extinct or has not been included in modern-day samples.

If the allele was segregating in an Asian population, it is improbable that all of the copies of 9RA in the Americas descend from more than one ancient sampling event from that population. It is unlikely that an allele at a frequency sufficiently low to destine it for extinction, or an allele the sole source of which is a small, geographically restricted population, would have been included in multiple migratory groups and maintained multiple times.

Haplogroup D
16223-16241-16301-16342
Entry time = 13-15 kya

Fig. 1. Map of the Americas that indicates the approximate locations of the On Your Knees Cave and related samples. Sample size (N) of one at each location unless otherwise noted.

Regional haplogroup frequencies in North America
(Fix, 2005)
The colonizing of the entire coast of North America (along with the Mississippi Valley) was accomplished in 120 generations, or some 3,000 years. This relatively rapid spread of the initial colonizing population would have deployed the founding mtDNA haplogroups to the farthest extent of the continent; A similar process would have extended colonization along the South American coast at roughly the same time. These pioneer coastal settlements, along with extensions up major river valleys such as the Mississippi, could have served as ‘staging areas’ (Anderson, 1990) for the continuing colonization of the continental interiors.

Haplogroup X in North and Central America

Is haplogroup X present in extant South American Indians?


Native American mtDNA RFLP haplotype phylogeny

Schurr TG, 2000, Mitochondrial DNA variation in Native Americans and Siberians, and its implications for the peopling of the New World, Amer Sci 88(3): 246-53

European populations with haplogroup X mtDNAs

Frequency ~2% per population

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Haplogroup X distribution in Eurasia

Haplogroup C distribution in Eurasia

mtDNA haplogroup frequencies in Siberia

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Haplogroup X network
Derenko, et al., (2001)

So how did X get to the Americas?

Prehistoric European migration to North America?

Clovis and Solutrean connection?
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Problems: Solutrean = Clovis

- Solutrean people not maritime adapted?
- Parallelism in manufacture of Clovis and Solutrean point?
- Solutrean culture in Iberia is several thousand years older than Clovis in Americas, leaving large time gap
- Why only haplogroup X brought to New World – when haplogroups H and U high at high frequency in Europeans (J, T, V also present in Iberia)?

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[Diagram showing genetic relationships among Circumarctic groups, including Aleuts, Yupik-Inupiaq-Inuit, and Native Americans.]

[Circumarctic groups are connected with lines indicating genetic flow or migrations.]

[Map showing post-glacial maximum population expansions across the Americas.]

[Map with expansion timelines: 6-9 kya, 13 kya, 4-7 kya, ~1 kya, 2-3 kya, ~0.5 kya.]

[Ancient vs. modern population diversity chart with bars representing different genetic groups and percentages.]

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Y chromosome haplogroup frequencies in the Americas
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Ages of NRY haplogroups

- M89 = 45,000 - 50,000 YBP
- M9 = 35,000 - 45,000 YBP
- M45 = 25,000 - 35,000 YBP
- M130 = 25,000 - 40,000 YBP
- M3 = 7,000 - 20,000 YBP
- M17 = 7,000 - 14,000 YBP
- M46 = 3,000 - 7,000 YBP

Abbreviated NRY SNP haplotype network

"Upper limit" on New World colonization = 15,000 ybp

Y chromosome SNP haplotype network

M130 [Haplogroup C]
M3 (and M45) [Haplogroup Q]
Table 3: Divergence Time Estimates with Standard Errors in Years

<table>
<thead>
<tr>
<th>Comparison</th>
<th>Upper bound by Tp with Y = 6</th>
<th>Lower bound by Tp with Y from Data</th>
<th>Mean from Tp</th>
</tr>
</thead>
<tbody>
<tr>
<td>Asia &gt; North Asia versus Native Americans</td>
<td>12,000 ± 4,000</td>
<td>14,700 ± 5,700</td>
<td>16,150 ± 5,700</td>
</tr>
</tbody>
</table>

Notes: The generation time was assumed to be 25 years with an effective Y-SN mutation rate = 0.0007 per generation. Standard errors of estimates obtained by jack-knifing over loci. (Wilcox 1996)

- Greenland Inuit: 60% European Y chromosomes, Norse influence

Zegura et al., 2004
Summary of genetic data

- Initial expansion into the New World appears to have occurred between 15,000-20,000 ybp.
- Most data support a single major expansion into the Americas giving rise to Amerindian populations, although still some questions about this viewpoint.
- Secondary expansions into North America by ancestral populations of Eskimo-Aleuts and Na-Dene Indians ~10,000 ybp.
- Patterns of regional divergence appearing in Americas with more data from various populations.
- Continuity of regional populations for ~6,000 years based on ancient DNA research.

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