

Admixture between Archaic and Modern Humans

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Fossils From Vindija Cave, Croatia



Hominin tooth from Denisova Cave, Altai Mtns, southern Siberia



Nucleotide site patterns

	Nucleotide Site Pattern		
	<i>ea</i>	<i>en</i>	<i>an</i>
Eur	1	1	0
Afr	1	0	1
Nea	0	1	1
Chmp	0	0	0
#	303,340	103,612	95,347

Ancestral allele (0) is shared with chimp.

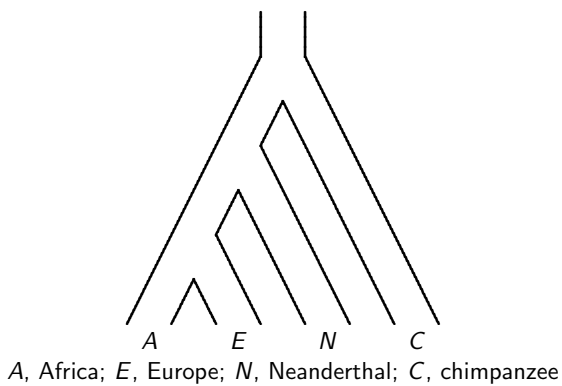
Mutant allele (1) shared by two human populations.

Pattern *ea*: most common; reflects history of population splits.

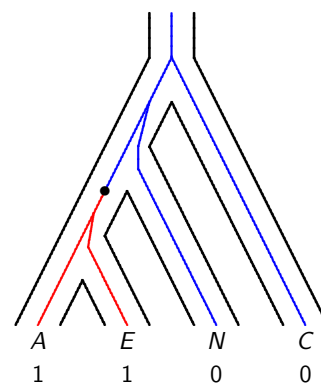
Patterns *en* & *an*: how do they arise?

Why does *en* exceed *an*?

Population tree

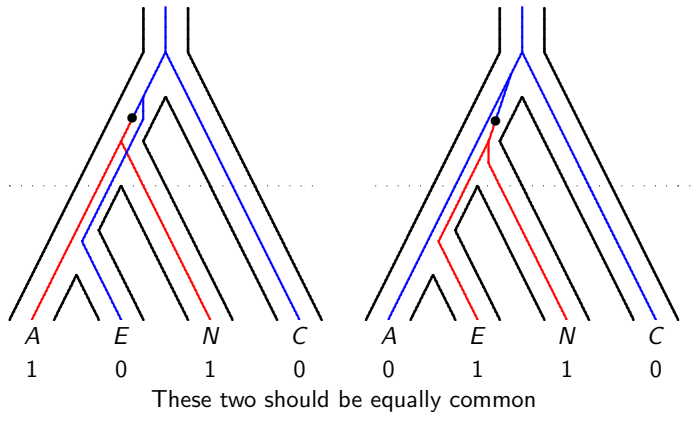


Embedded gene genealogy with mutation



- ▶ Genealogy of 4 genes shown in color.
- ▶ Bullet (●) marks mutation from allele 0 to allele 1.
- ▶ Descendants of mutant have allele 1; others have 0.
- ▶ Gene genealogy matches phylogeny
- ▶ Mutant allele shared by closest relatives, A and E.

Lineage sorting

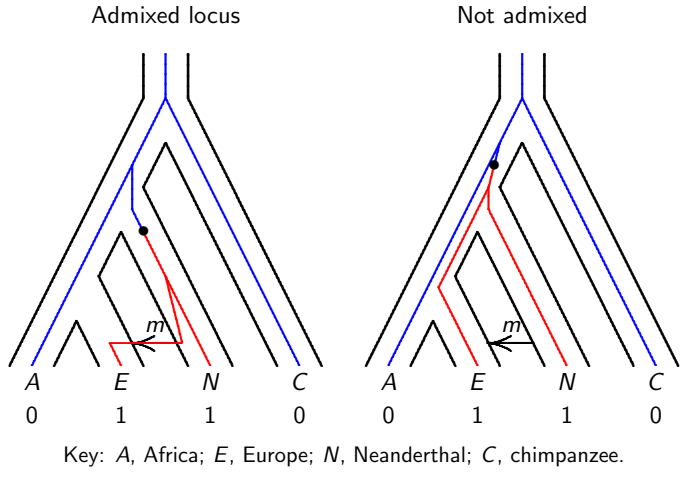


Nucleotide site patterns again

	Nucleotide Site Pattern		
	<i>ea</i>	<i>en</i>	<i>an</i>
European	1	1	0
African	1	0	1
Neanderthal	0	1	1
Chimpanzee	0	0	0
# sites	303,340	103,612	95,347

Common pattern (*ea*) reflects history of population splits.
 Absent admixture, the other two should be equally common
 Why does *en* exceed *an*?

Neanderthal admixture inflates *en* site pattern



Estimate from Neanderthal DNA

- ▶ DNA of modern Europeans is 1.3–2.7% Neanderthal.
- ▶ Same is true for modern people of east Asia and Papua New Guinea, but not Africa. Green et al (2010)
- ▶ Admixture must have occurred *after* moderns left Africa but *before* they expanded throughout the world.

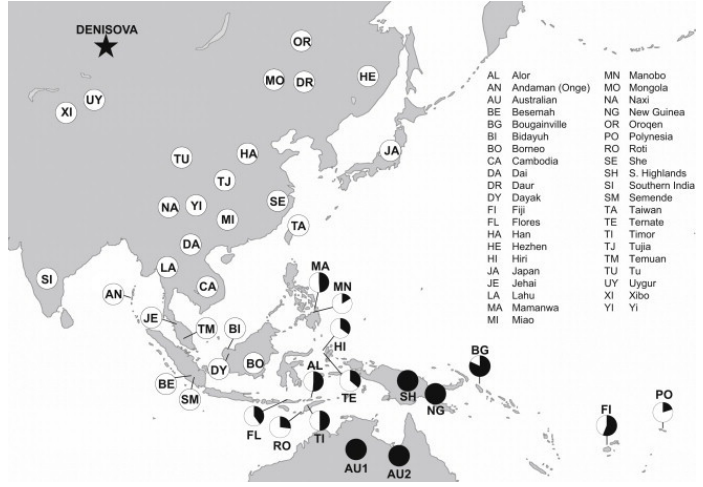
Eurians share some derived alleles with archaics

Neanderthal matches French 4.6% more often than Yoruban (African).
 Denisova matches French 1.8% more often than Yoruban (African).
 Archaic component of Eurasian genome more Neanderthal than Denisovan.
Green et al 2010; Reich et al. 2010.

So do Asians and Papuans

Asians and Papuans carry as many Neanderthal alleles as Europeans do: 1.3–2.7%.

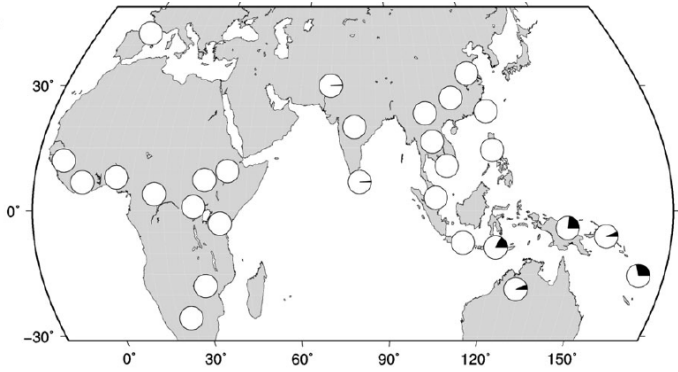
Denisovan DNA most common in Australia, NG, and Oceania



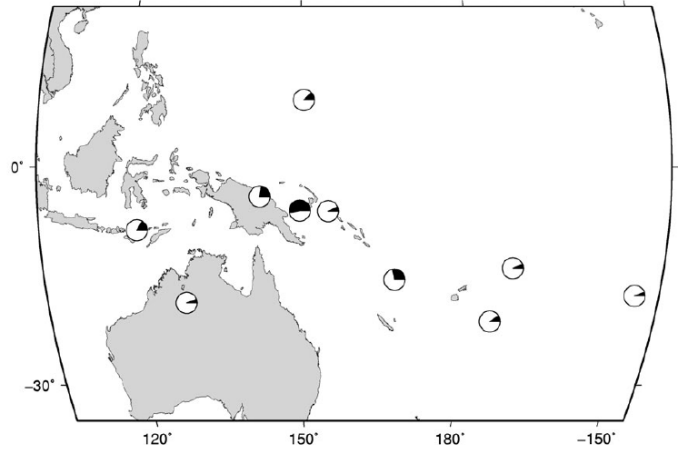
OAS1 innate immunity locus

- ▶ two forms of gene in Melanesia:
- ▶ one shared with rest of world
- ▶ one only in Melanesia

Worldwide frequency of Melanesian OAS1 allele



Melanesian OAS1 allele w/i Melanesia



Melanesian OAS1 allele is old yet young

- ▶ The 2 alleles differ at many nucleotide sites ⇒ separation time 3.4 my.
 - ▶ Long (90 kb) LD block ⇒ they've been together only 25 ky
 - ▶ Melanesian allele matches that in Denisovan hominin skeleton.
- ⇒ archaic admixture into Melanesia

Summary

- ▶ Our ancestors expanded out of Africa 50 kya, and mated with surrounding archaic populations.
- ▶ Yet only a few archaic genes entered the modern population.
- ▶ Europeans are part Neandertal, and Melasians are part Denisovan.