Semester Wrapup

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Local isolation versus gene flow

1. Migration among savannah foragers and among tropical horticulturalists. What was it like in the past?
2. Fine-scale geographic structure in Britain
3. Decay of IBD with distance in Europe
4. Indian castes
5. Population structure in pre-agricultural Africa.
6. Denisovan subpopulations
7. Large effective size of the superarchaic population.

Migration among Ju/'hoansi communities (Harpending)

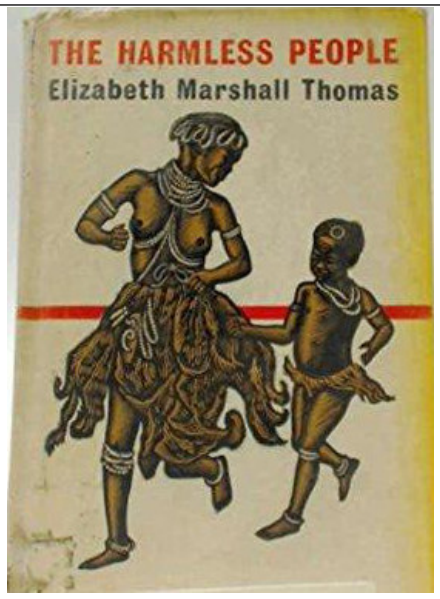
Birthplace	Adult Residence								
	A	B	C	D	E	F	G	H	I
A	45	2	0	0	0	3	9	0	9
B	6	16	14	0	5	1	2	1	14
C	7	8	40	5	7	3	1	1	1
D	0	5	10	19	0	2	0	3	0
E	6	0	0	0	40	13	2	0	3
F	14	10	5	2	10	40	3	0	1
G	10	0	0	0	1	2	48	0	1
H	3	1	2	9	1	2	0	29	2
I	1	3	2	0	0	0	0	0	9

Savannah foragers; high gene flow ⇒ small group differences.

Migration on Bougainville Island (Friedlaender)

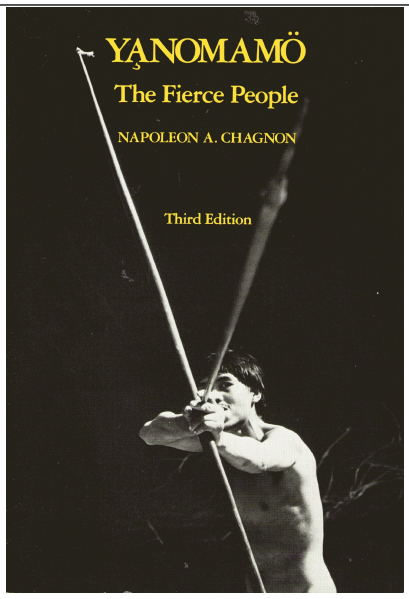
Birthplace	Adult Residence															
	A	B	C	D	E	F	G	H	I	J	K	L	M	N	O	P	Q
A	48	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0
B	0	8	2	0	0	0	0	0	0	0	0	0	0	0	0	0	0
C	0	0	112	1	0	0	0	0	0	0	0	0	0	0	0	0	0
D	0	0	5	23	5	0	0	0	0	0	0	0	0	0	0	0	0
E	0	0	1	6	57	0	0	1	0	0	0	0	0	0	0	0	0
F	0	0	1	1	0	34	13	2	0	0	0	0	0	0	0	0	0
G	0	0	0	0	0	1	1	1	0	3	0	0	0	0	0	0	0
H	0	0	1	0	0	6	2	37	15	0	0	1	0	0	0	0	0
I	0	0	0	0	0	0	0	4	49	1	0	0	0	0	0	1	1
J	0	0	0	0	0	0	0	4	4	89	0	0	2	0	0	0	0
K	0	0	0	0	0	0	0	0	0	4	16	2	0	0	0	0	1
L	0	0	0	0	0	0	0	0	0	1	4	35	4	0	0	1	0
M	0	0	0	0	0	0	0	0	0	0	0	0	4	0	0	0	0
N	0	0	0	0	0	0	0	0	0	0	0	0	0	32	0	0	0
O	0	0	0	0	0	0	0	0	0	0	0	0	0	1	47	0	0
P	0	0	0	0	0	0	0	0	0	0	0	0	1	0	0	35	0
Q	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1	101

Tropical forest horticulturalists; low gene flow ⇒ large group differences.



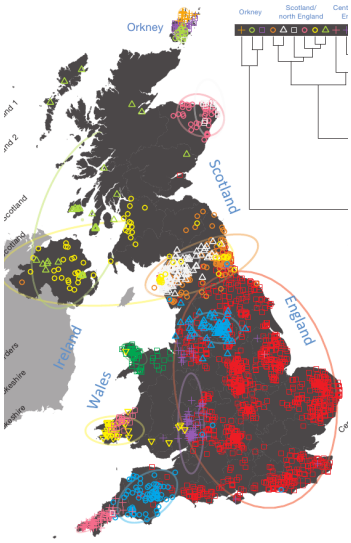
Peaceful foragers

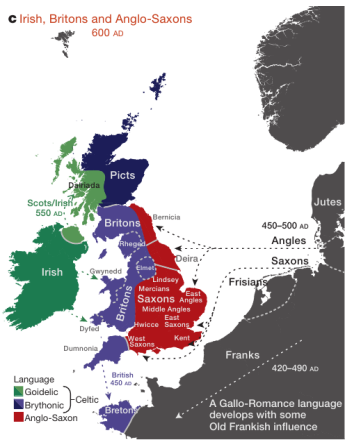
Ethnography described peaceful foragers.



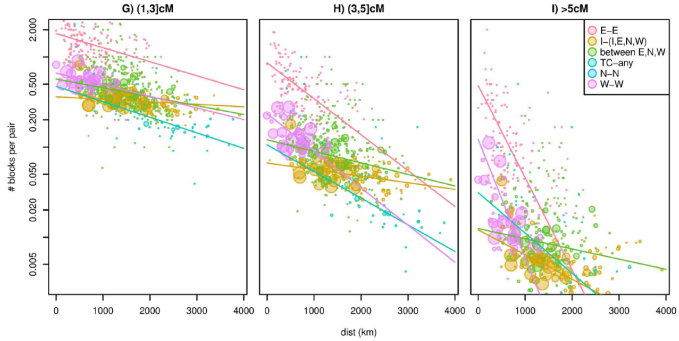
Warlike horticulturalists

It also described warlike horticulturalists.





Geographic decay of recent relatedness



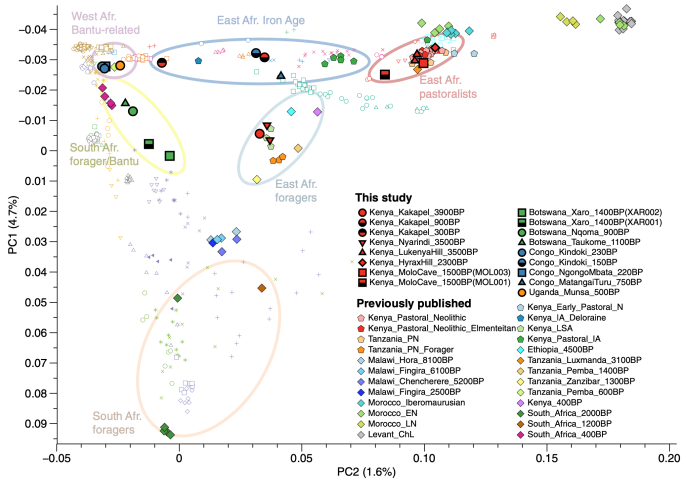
Genetic similarity versus geographic distance. Small dots are pairs of individuals. E, Eastern Europe; W, Western Europe; N, Northern Europe; I, Italy & Iberia; TC, Turkey & Cyprus. (Ralph & Coop 2013)

Admixture and social class in India

Groups with higher social class have more ANI admixture.

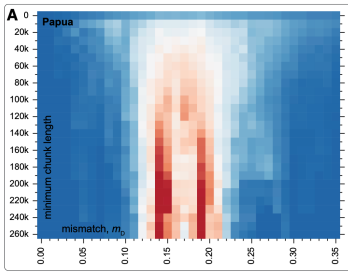
Reich (2018) argues that these groups are at least 4000 years old and have retained distinctive genetics since then.

Irawati Karve suggests that the caste system began when a ruling elite imposed itself upon what had been a tribal society. The tribes became *jati* (groups w/i the Indian caste system), and were organized for labor.



Large genetic differences btw ancient African foragers (Wang et al, 2017).

Papuans got DNA from 2 Denisovan pops



Jacobs et al (2019)

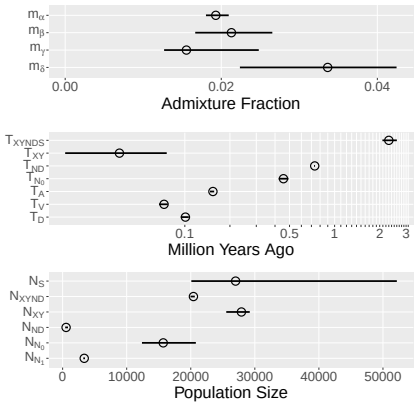
Vertical axis: length of introgressed segment

Horizontal: diff btw segment and Denisovan genome as fraction of Denisovan-modern diff.

Color: number of fragments in this bin of length × dissimilarity

Two Denisovan populations: one 0.15 and one at 0.2.

Parameter estimates



Superarchaic population separated ∼2 mya. It was large—between 20,000 and 50,000—or deeply subdivided.

neandersovan population (N_{ND}) was tiny, and split early ($T_{ND} > 700$ kya) to form Neanderthals and Denisovans.

∼3% admixture into neandersovans from superarchaics.

Conclusion about local isolation versus gene flow

The comparison between Ju/'hoan and Bougainville data suggested that forager populations were less isolated and more peaceful.

Yet there seems to be plenty of geographic variation in ancient allele frequencies. So perhaps the Ju/'hoansi are not representative of the human past.

Invasions and co-existence (or not)

1. Pots or people? V. Gordon Childe vs. Grahame Clark
2. The European Neolithic was a movement of people.
3. Two patterns: (1) long-term co-existence w/ gene flow from forager → farmer; (2) near-total replacement.
4. India: co-existence and gene flow
5. Africa: both patterns

Childe: one population replaces another, over and over

> *We find certain types of remains—pots, implements, ornaments, burial rites and house forms—constantly recurring together. Such a complex of regularly associated traits we shall call a "cultural group" or just a "culture." We assume that such a complex is the material expression of what today we would call "a people."*
>
> *—1929*

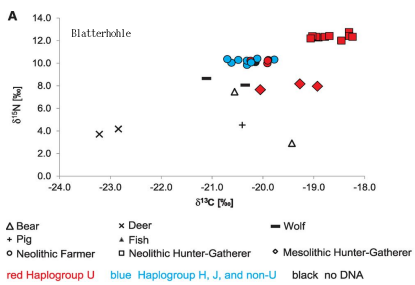
In particular, Childe saw the European Neolithic as a movement of farming peoples into Europe, replacing the resident foragers.

European prehistory after 1960

Grahame Clark

- Agriculture spread slowly across Europe—not an invasion.
- Diffusion of an idea—movement of pots, not people.
- Archaeology developed a deep skepticism toward explanations that involved large movements of people.

The Blätterhöhle site in Germany



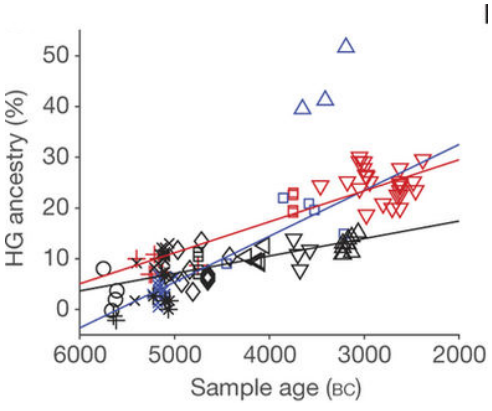
Red diamonds: Mesolithic foragers

Red squares: Neolithic (aged) fishers with Mesolithic DNA.

Blue and red circles: Neolithic farmers w/ Middle-Eastern DNA.

Foragers and farmers lived side by side, with some gene flow from forager to farmer.

Mesolithic ancestry increases with time

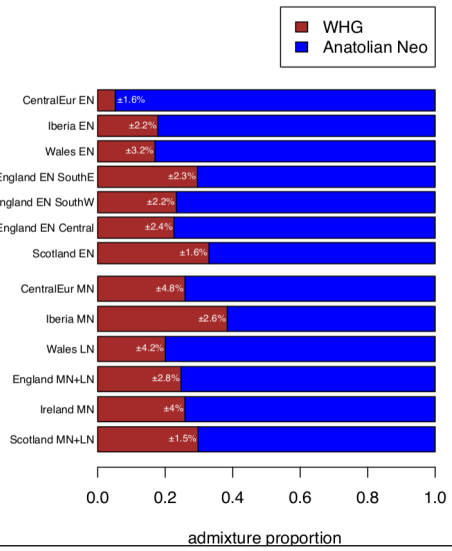


Farmers and foragers co-existed and interbred over millennia.

△ admixture greatest in Germany

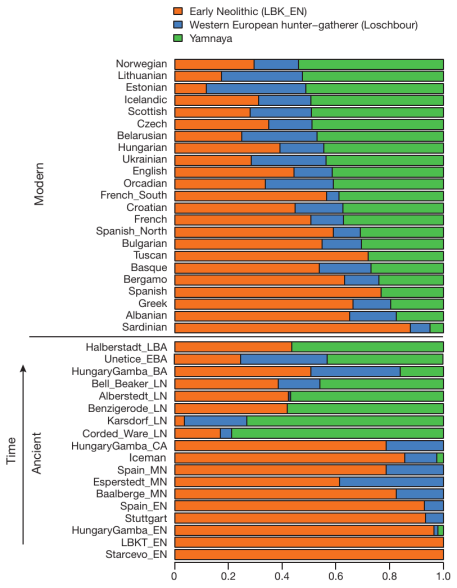
Lipson et al. (2017)

Mesolithic admixture in Neolithic Britain



WHG fraction doesn't increase with time.

Neolithic Brits didn't interbreed with foragers.



Corded ware: 3/4 Yamnaya in autosomal DNA

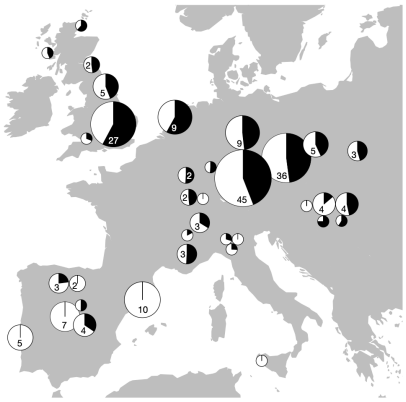
Yamnaya fraction even larger for yDNA

Most mtDNA came from the Middle East.

Yamnaya fraction declines over time as invaders interbreed.

Haak et al (2015)

Fraction of steppe ancestry w/i Bell Beaker genomes



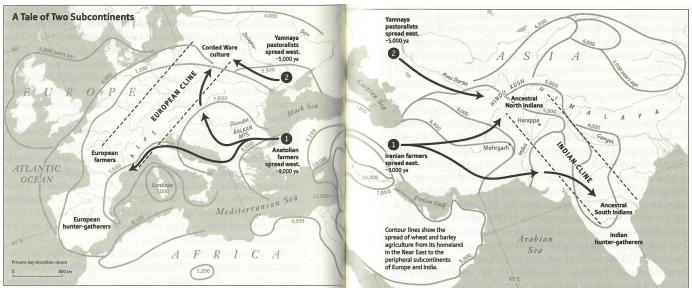
Substantial steppe ancestry everywhere except Iberia.

Within Iberia, France, and Britain, the non-steppe component is most similar to earlier Iberian genomes.

Suggests Bell Beaker spread N from Iberia without much mixing.

(Olalde 2018)

European and Indian clines



In Europe, Yamnaya ancestry declines from NE to SW.

In S Asia, it declines from NW to SE.

The stories of the two subcontinents are very similar.

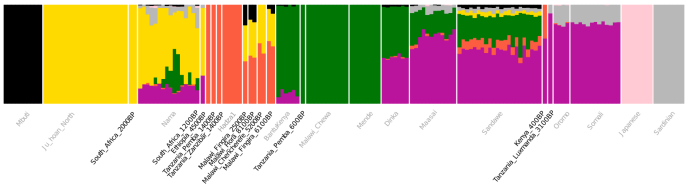
Admixture and social class

Groups with higher social class have more ANI admixture.

Irawati Karve suggests that the caste system began when a ruling elite imposed itself upon what had been a tribal society. The tribes became *jati* (groups w/i the Indian caste system), and were organized for labor.

Did this also happen in Europe? We don't know. In India, the caste system reduced gene flow, so the ancient pattern has been preserved.

African admixture



Yellow: Ju/'hoansi; Orange: Hadza; Black, pygmy.

Other populations show varying levels of admixture with these.

These show no admixture. (Later studies modify this conclusion.)

One-way gene flow from foragers to pastoralists and agriculturalists.

The African pattern of one-way gene flow from foragers → agriculturalists is reminiscent of the Blätterhöhle site of the German Neolithic.

Africa is unique: it provides an ethnographic record of these interactions.

Interbreeding between farmers and foragers

In some parts of Africa, farmers interbred with foragers.

In Malawi, the modern population contains no trace of the ancient foragers who lived there.

Conclusions about invasions and co-existence

Genetic evidence documents numerous territorial invasions on several continents: the European Neolithic, the Yamnaya, who invaded both Europe and India early in the Bronze age, the Bantu of Africa, etc.

In some cases, invaders co-existed with previous occupants, with limited gene flow over thousands of years.

In other cases, invaders replaced previous occupants almost completely.

Y chromosomes are often replaced more completely than mtDNA, implying that invading males have greater reproductive success than native ones.

Adaptive evolution

1. There is no truth to the old view that adaptive evolution has stalled in the human species.
2. Adaptive responses to Neolithic diets.
3. Maladaptive side-effects.
4. Adaptive introgression from archaic hominins.

Conventional wisdom

Something must have happened to weaken the selective pressure drastically. We cannot escape the conclusion that man's evolution towards manness suddenly came to a halt.

—Ernst Mayr 1963

Natural selection has almost become irrelevant in human evolution. There's been no biological change in humans in 40,000 or 50,000 years. Everything we call culture and civilization we've built with the same body and brain.

—Stephen Jay Gould 2000

Certainly, human nature is fixed. It's universal and unchanging —common to every baby that's born, down through the history of our species.

—Helena Cronin 2000

DNA sequences from region of human lactase gene

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     cgcttcaggcattcctatctaaacagaccaacgtaAgggtacaatgcctaacccagacgtttcaactct
20   ..........................................................................
21   ..........................................................................
22   ..........................................................................
23   ..........................................................................
24   ..........................................................................
25   ..........................................................................
26   ..........................................................................
27   ..................t.......................................................
28   ..................t.......................................................
29   ..................................................c.......................
37   ...................................G..a.gt.....t.........gac.c.tgtct.
38   ...ccgga....gat..at..gg..c.....tc.gGaaa.g..ccttt...tg......c...t.t...
39   ...ccgga....gat..at..gg..c.....tc.gGaaa.g..ccttt...tg......c...t.t...
40   ..tcc...agtag.t.cat..g.....t..ttccgG..a.gt.....t.........gac.c.tgtct.
41   ..tcc...agtag.t.cat..g.....t.gttccgG..a.gt.....t.........gac.c.tgtct.
42   ..tcc...agtag.t.cat..g.....t.gttccgG..a.gt.....t.........gac.c.tgtct.
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44   ..tcc...agtag.t.cat..g.....t..ttc.gG..acgt.....t.........gac.c.tgtct.
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46   ...ccgga....gat..at..gg..c.....tc.gGaaa.g..ccttt...tg......cg.gt.t..c
47   ..tcc...agtag.t.cat..g.....t.gttccgG..a.gt.....t.........gac.c.tgtct.
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49   ..tcc...agtag.t.cat..g.....t.gttccgG..a.gt.....t.........gac.c.tgtct.
50   tatccgga....g.tc.atcgg.tc.g.tg.tc.gG..a.g.g....tg....ggt...cg.gt.t..c
51   ta.ccgga....g.t..atcgg.tc.g.tg.tc.gG..a.g.g....tg....ggt...cg.gt.t..c
52   ta.ccgga....g.t..atc.g.tc.g.tg.tc.gG..a.g.g....tg....ggt...cg.gt.t..c
53   ta.ccgga....g.t..atcgg.tc.g.tg.tc.gG..a.g.g....tg....ggt...cg.gt.t..c
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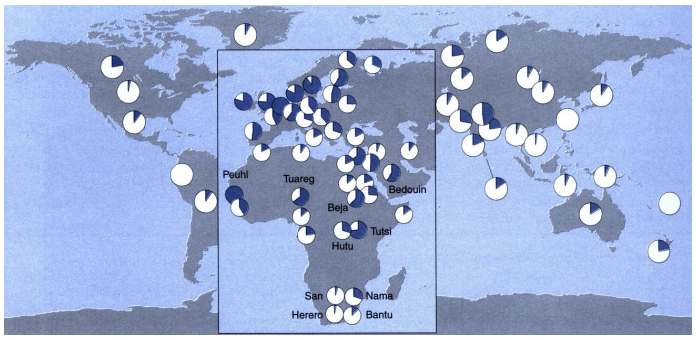
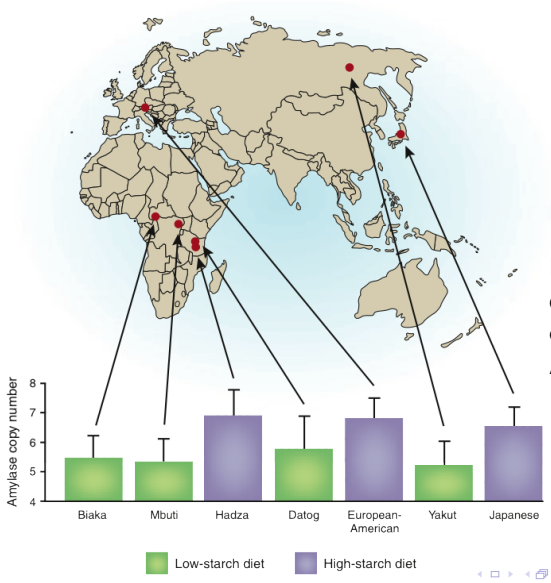
Distribution of lactase persistence (dark blue)

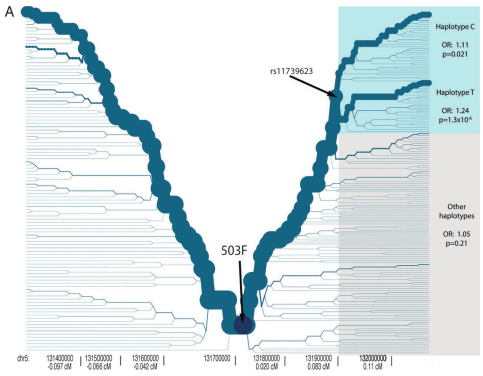




Population diffs

High-starch diet ⇒ more copies of AMY1

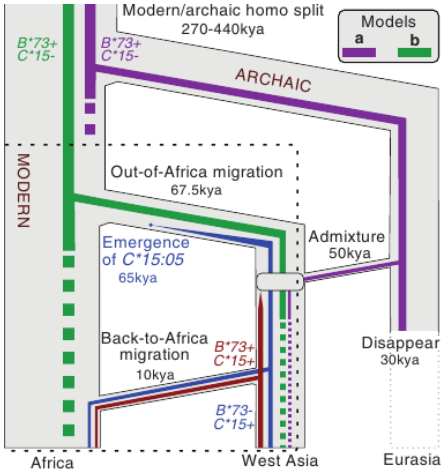
Crohn's disease caused by harmful hitch-hiker



Blue: no recombination. Assoc. btw harmful hitch-hiker at 503F & Crohn's.

Gray: recombination separates harmful hitch-hiker. No assoc. w/ Crohn's.

History of B*73:01-C*15 HLA haplotype



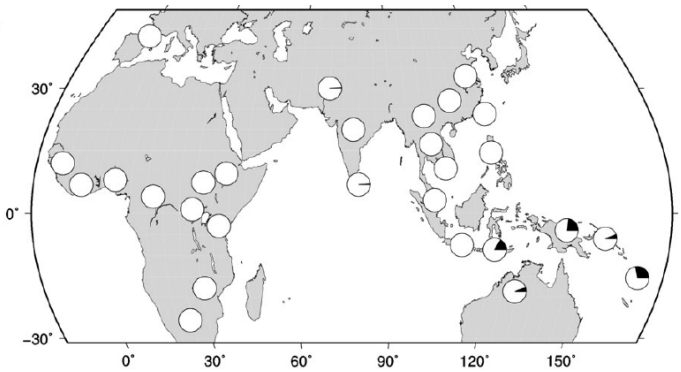
Other HLA alleles

There are other HLA alleles with similar stories.

Abi-Rached et al (2011) estimate that $> 50\%$ of Eurasian HLA alleles came from archaics.

Archaics contributed a lot to the adaptive immune systems of modern humans.

Worldwide frequency of Melanesian OAS1 allele



Melanesian OAS1 allele is old yet young

- The 2 alleles differ at many nucleotide sites $\Rightarrow$ separation time $\sim$3.4 my.
- Long (90 kb) LD block $\Rightarrow$ they've been together only $\sim$25 ky
- Melanesian allele matches that in Denisovan hominin skeleton.

$\Rightarrow$ archaic admixture into Melanesia

Conclusions about adaptive evolution

There is abundant evolution for recent adaptive evolution in humans.

Lactase persistence has evolved multiple times in pastoral populations.

Copy number of an amylase gene increases in response to high-starch diets.

Crohn's disease seems to be a maladaptive side effect of selection for an advantageous allele in Europe.

Many alleles related to immunity were acquired from archaics.