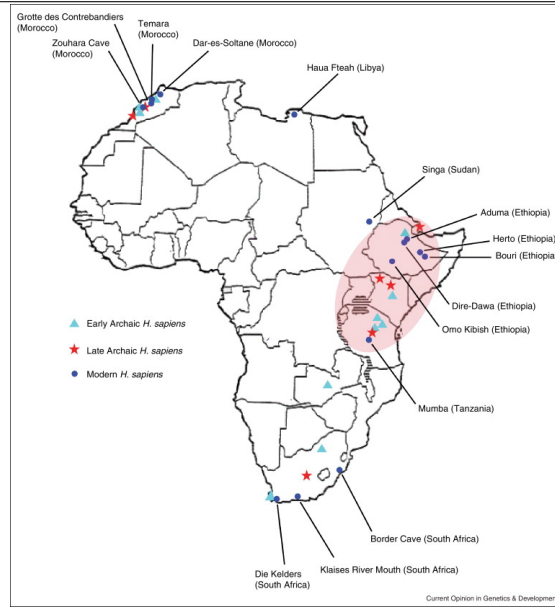


Lactase Persistence (LP) in Africa

Alan R. Rogers

March 30, 2016



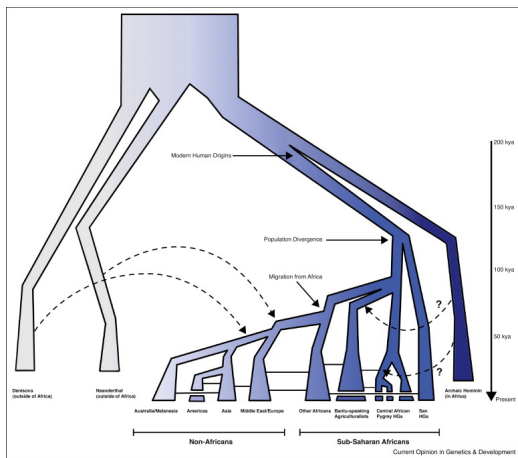
Early modern humans in Africa

Pigment, art, shell beads: 164 kya (S Africa); 82 kya (N Africa)

Microlith stone tools: 71 kya (S Africa)

Current Opinion in Genetics & Development

Population tree



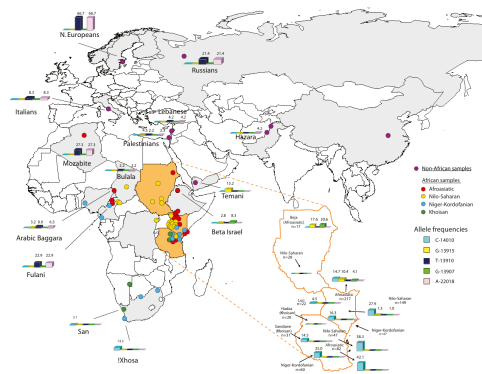
Deep split inside Africa

African archaics → Africans

Denisovan and Neanderthal → Eurasians

Current Opinion in Genetics & Development

Geographic distribution of LP alleles



Sample: 819 Africans & 154 non-Africans

5 LP alleles

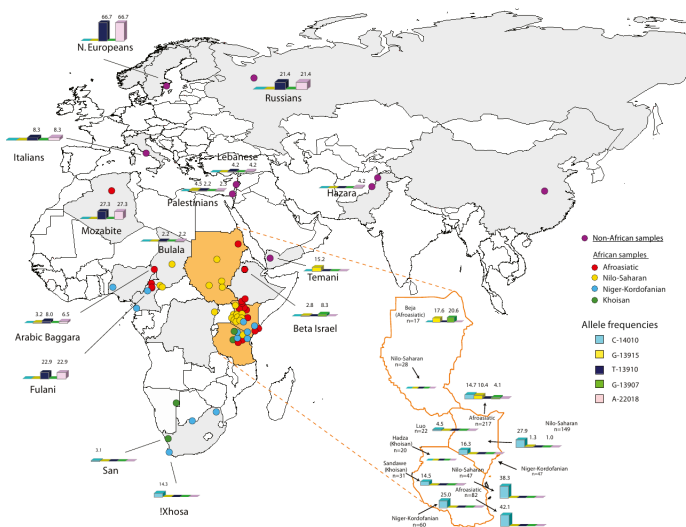
T13910, A22018: N Europe, Sahara

C14010, G13907: E Africa

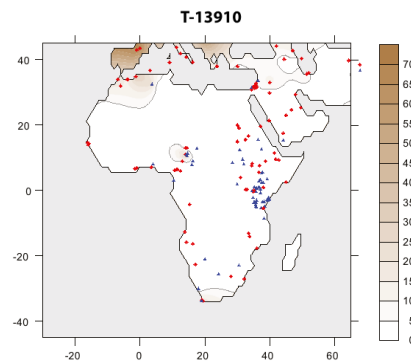
G13915: E Africa & Arabia

Sprinkling of LP alleles in S and W Africa.

Ranciaro et al (2014)



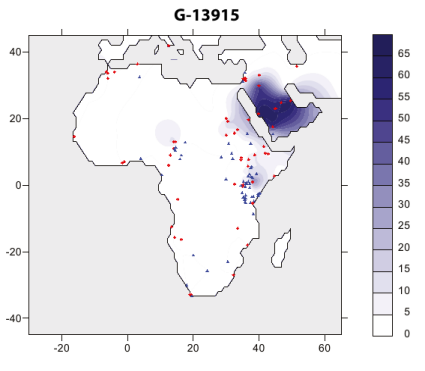
European LP allele



Most common in Europe

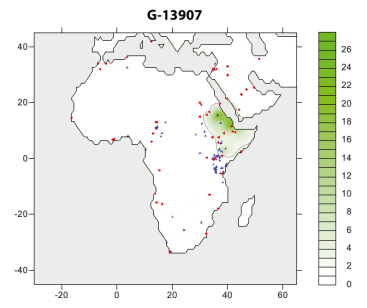
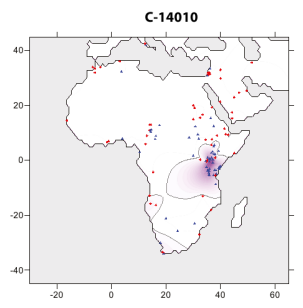
Also NW Africa & Nigeria (the Fulani)

Arabian LP allele

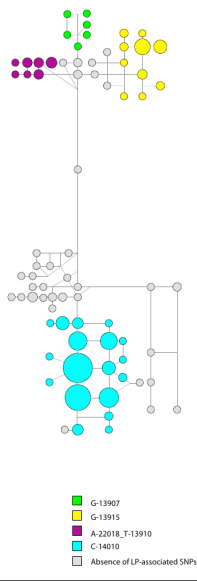


Most common in Arabia
Also E and C Africa

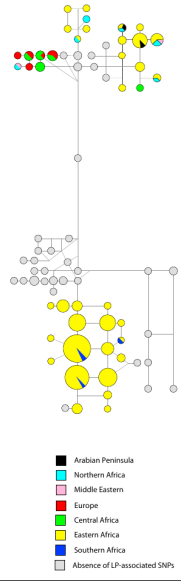
E African LP alleles



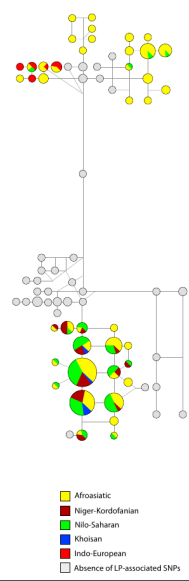
A Genotype



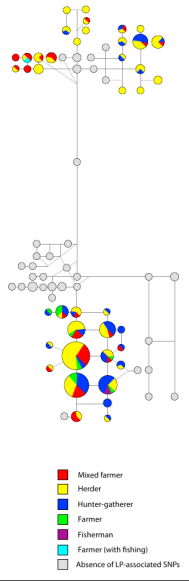
C Geographic Regions



B Language Family

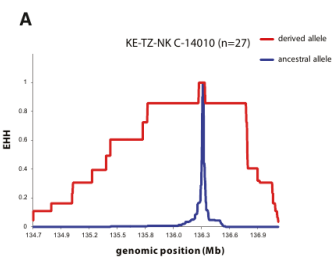


D Subsistence Pattern



Evidence for selection at C14010

Predominantly E African allele in Kenya & Tanzania



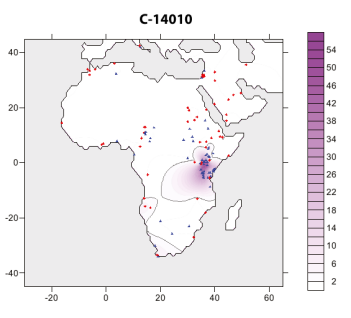
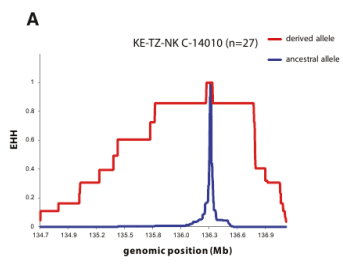
Extend Haplotype Homozygosity (EHH) extends for ~2 Mb (mega bases) in derived allele.

EHH extends only a little way in ancestral allele.

Strong evidence for recent selection.

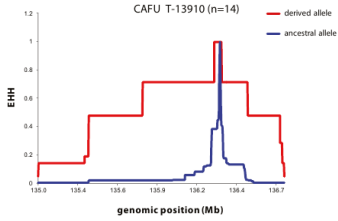
Evidence for selection at C14010

Predominantly E African allele in Kenya & Tanzania



Evidence for selection at T13910

Predominantly European allele among Fulani



Extend Haplotype Homozygosity (EHH) extends for 1.7 Mb in derived allele.

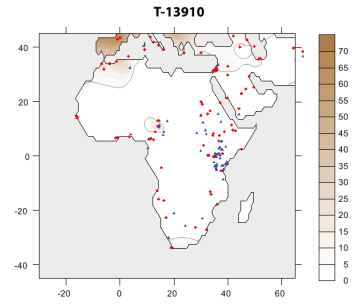
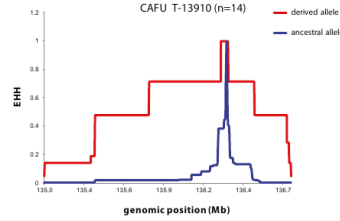
Only about 1 Mb in Europe.

EHH extends only a little way in ancestral allele.

Strong evidence for recent selection.

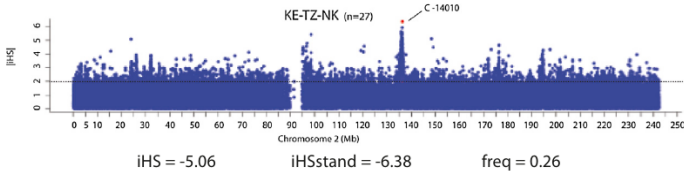
Evidence for selection at T13910

Predominantly European allele among Fulani

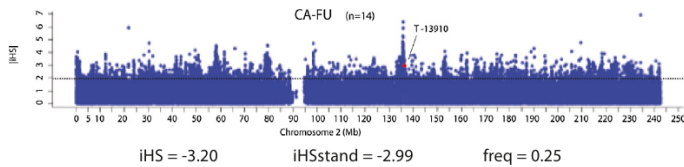


LD scan showing two SNPs associated with LP

E Africa



Fulani



Soft sweep in E Africa?

Hard selective sweep Begins from a single copy of an advantageous mutation. Easy to detect with LD.

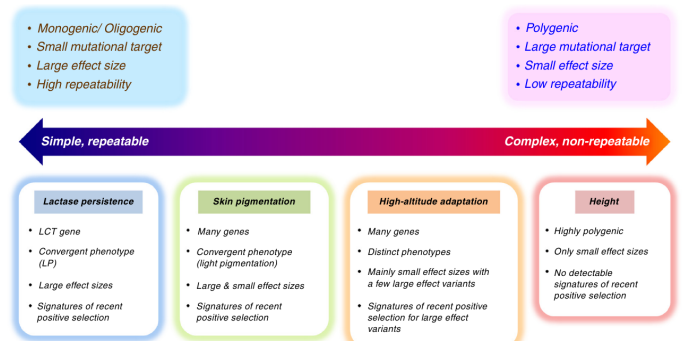
Soft selective sweep Begins from multiple copies, possibly of multiple alleles with similar advantageous functions. Hard to detect.

Existence of several LP alleles in E Africa suggests a soft sweep. May explain absence of evidence for selection for some alleles.

Missing LP alleles

Some LP individuals lack all known LP alleles. Includes 47% of Hadza, a foraging population with no history of dairying. Lactase also breaks down phlorizin, a bitter substance used to treat malaria. Do the Hadza have a history of exposure to phlorizin?

Modes of adaptation



Current Opinion in Genetics & Development

Jeong & Di Rienzo (2014)

Summary

1. Africa: home of modern humans; high genetic diversity; deep population tree
2. Has 5 LP alleles.
3. Selective sweep at C14010 in E Africa
4. Also at T13910 (European allele) among Fulani
5. No sweep at other 3 alleles.
6. Possible soft sweep.
7. Some Africans have LP but lack all known alleles.