

Evolution of Lactase Persistence

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Outline

- ▶ Lactose, lactase, and lactase persistence
- ▶ Evidence of a selective sweep
- ▶ Prehistory of Europe

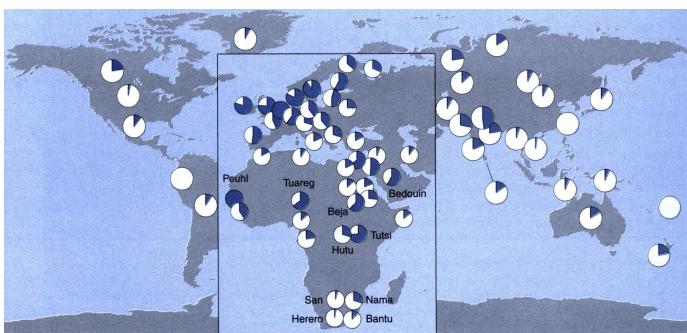
The trouble with fresh milk

- ▶ Contains the sugar *lactose*
- ▶ Digesting lactose requires the enzyme *lactase*
- ▶ Most humans don't produce it after age 5.
- ▶ Fresh milk gives them gas and diarrhea.
- ▶ 8000 years ago, all humans had this problem.

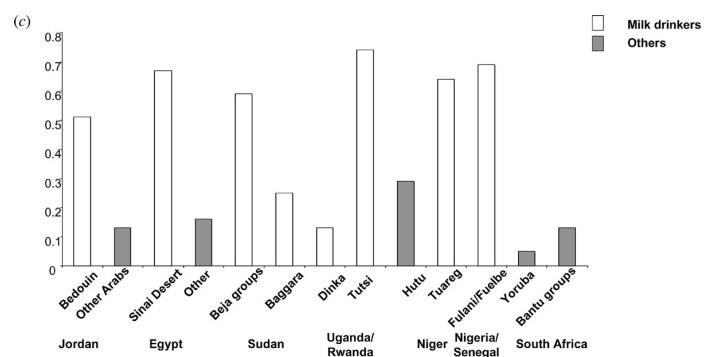
Lactase persistence

- ▶ Some modern humans produce lactase throughout life.
- ▶ Digest fresh milk as adults.
- ▶ Caused by mutation near lactase gene.
- ▶ When and where?

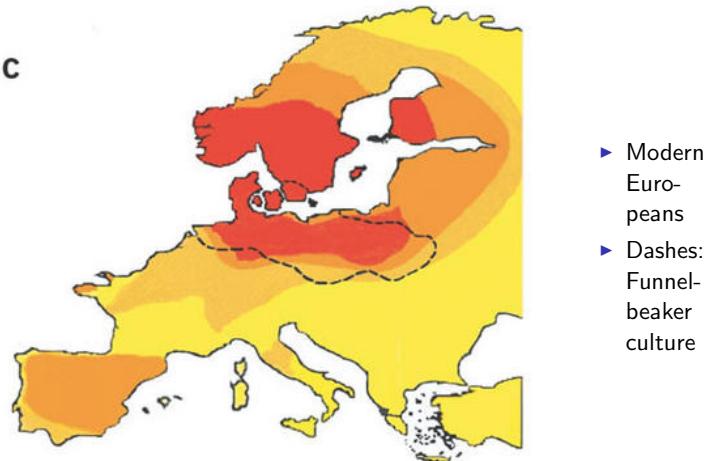
Distribution of lactase persistence (dark blue)



Within countries, lactase persistence more common in populations that drink milk



Lactase persistence in Europe



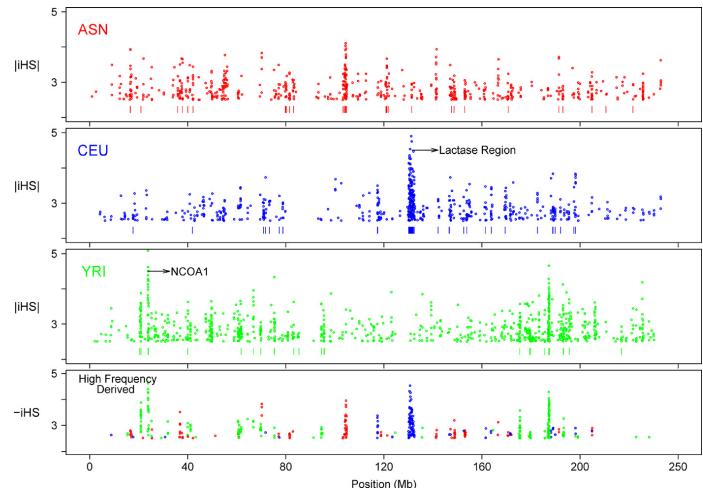
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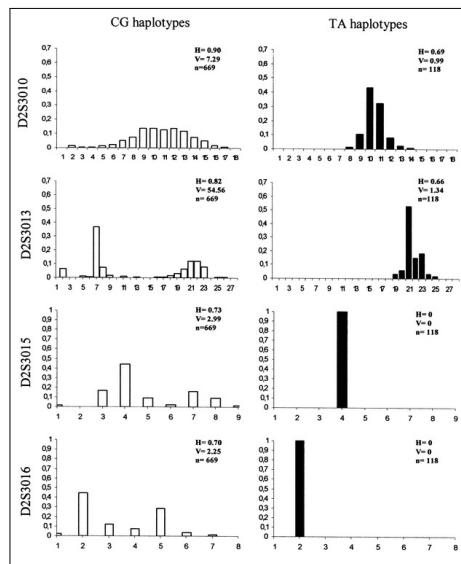
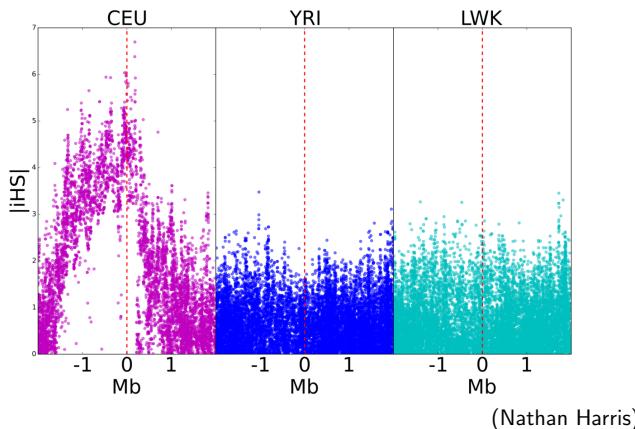
Evidence for a selective sweep

- ▶ In Europeans, persistence allele surrounded by a million bases of LD.
- ▶ Indicates strong selection.
- ▶ Statistical tests reject the drift hypothesis (Bersaglieri et al 2004)
- ▶ Increasing for ~10,000 years (Coelho et al 2005).

LD surrounds lactase gene in Europe



Huge block of LD around lactase allele in Europe

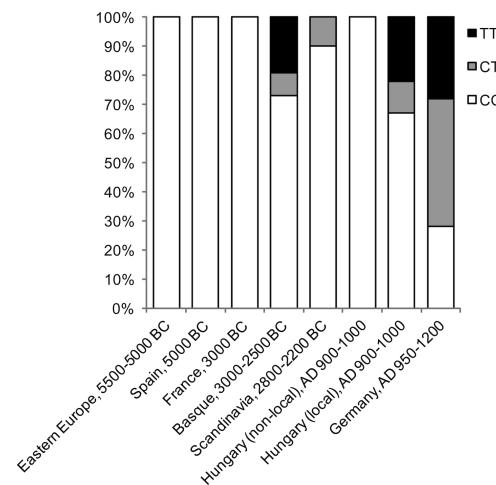


- ▶ Rows are different STRs
- ▶ Lactase persistence allele: haplotype TA.
- ▶ Has reduced SNP variation,
- ▶ Indicates recent origin.
- ▶ Age: 7,450 or 12,300 years (depending on assumptions)

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Lactase persistence in ancient Europeans

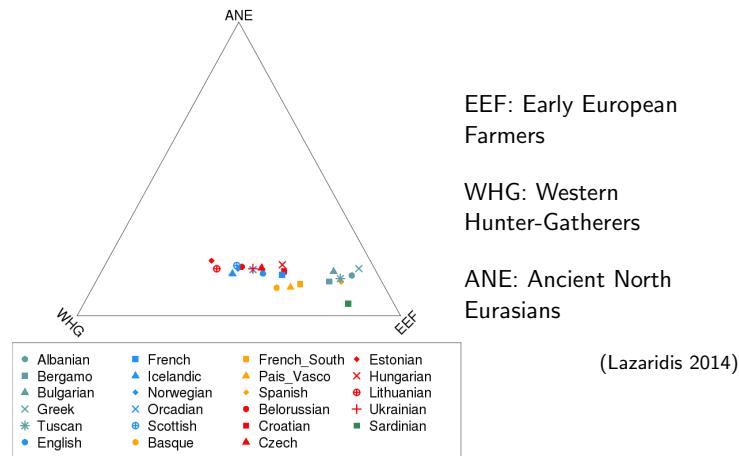


T is lactase persistence allele

Appeared in Europe by 2500 BC
(Krüttli et al 2014)

What was happening there 5000 y ago?

Modern Europeans derive from 3 ancestors

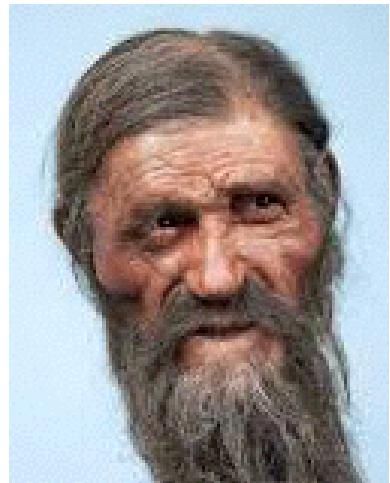


Old Europe



- ▶ Expanded into Europe from Middle-East and Anatolia beginning 7000 bp.
- ▶ Earliest European farmers.
- ▶ Lactose intolerant.

Ötzi, the Iceman



- ▶ 3300 BC on border between Italy and Austria
- ▶ Hair had high levels of copper and arsenic—a metal smith.
- ▶ Last meals: chamois and red deer meat, and einkorn wheat
- ▶ Lactose intolerant.

Long house



Sculpture



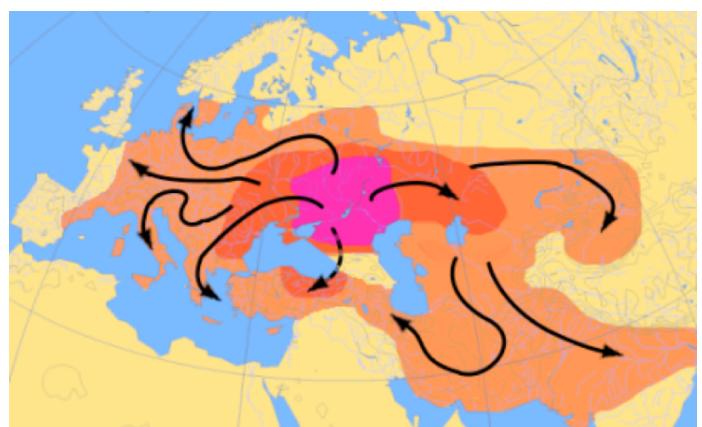
The Yamna culture: pastoralists



Mare's milk

- ▶ One kg of mare's milk has 190 Calories of fat and protein 250 Calories of lactose.
- ▶ With 5 kg per day yield one mare feeds two lactase persistent children.
- ▶ Less than one non-lactase-persistent child.
- ▶ Lactase persistence more than doubles food supply with one simple mutation.
- ▶ This advantage cannot be appropriated by others.

Indo-European expansion



Indo-European languages



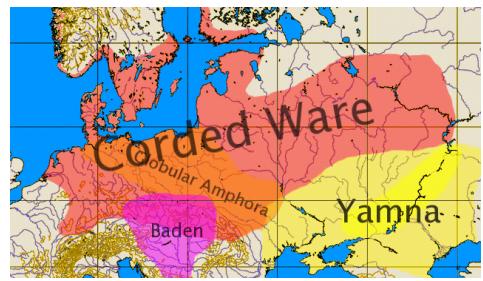
Tocharian



Modern Uighurs



Corded ware culture

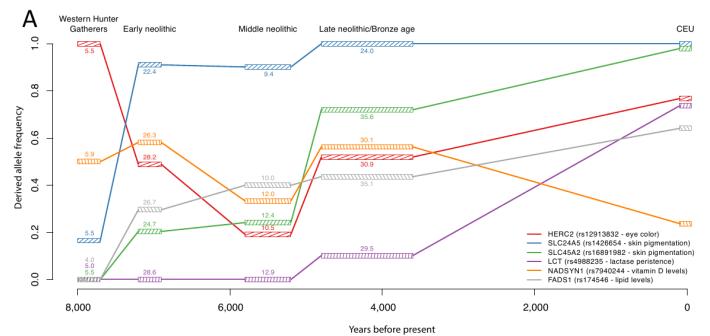


2400 BC
Overran Europe
Possibly introduced Indo-European languages
And maybe also lactase persistence

Study of Mathieson et al 2015

- ▶ DNA from 83 ancient Europeans.
- ▶ Track changes in allele frequencies over time.

History of evolution in Europe



Lactase persistence begins in Europe around 4000 BP.

Summary

- ▶ Recent adaptive evolution in lactase persistence.
- ▶ Appears in Europe about 5000 y ago.
- ▶ May have arrived with Indo-Europeans.