Evolution of Lactase Persistence

Alan R. Rogers

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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
▶ Digested 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

- 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)

Huge block of LD around lactase allele in Europe

(Nathan Harris)
Lactose, lactase, and lactase persistence

Evidence of a selective sweep

Prehistory of Europe

Lactase persistence in ancient Europeans

This 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

EEF: Early European Farmers

WHG: Western Hunter-Gatherers

ANE: Ancient North Eurasians

(Lazaridis 2014)

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 mean, and einkorn wheat

Lactose intolerant.

Long house

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 mean, and einkorn wheat

Lactose intolerant.
Mare's milk

- One kg of mare's milk has 190 Calories of fat and protein and 250 Calories of lactose.
- With 5 kg per day yield one mare feeds two lactase persistent children.
- Less that 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.