

LACTASE PERSISTENCE

For decades, lactase persistence has been the textbook example of gene–culture coevolution: when humans started herding cattle and drinking milk, the ability to digest lactose in adulthood supposedly became a major nutritional advantage, and natural selection drove the relevant variants to high frequency. Indeed, the region around LCT shows one of the strongest signals of positive selection in the human genome, with extreme extended homozygosity. See Schlebusch et al. (1)

In recent years, however, this simple story has been questioned. Archaeological and genetic evidence showed that people were using milk thousands of years before lactase persistence became common, and some authors suggested that selection at the LCT locus might have been driven by something else (for instance immune-related genes in the same region), rather than by milk consumption itself.

A new large-scale study published in bioRxiv (2) focusing on South Asia brings the story back into sharp focus. By analysing ~8,000 present-day and ancient genomes from India, Pakistan and Bangladesh, the authors show that the classic Eurasian lactase persistence allele (-13.910*T) arrived in South Asia via Steppe pastoralist ancestry. In most South Asian populations, its present-day frequency is explained almost entirely by ancestry, not by local selection.

But two striking exceptions stand out: the Toda in South India and the Gujjar in Pakistan, both traditional pastoralist groups whose subsistence has long revolved around milk and dairy products. In these two populations, the lactase persistence allele reaches frequencies comparable to Northern Europe and shows clear, independent evidence of very strong recent positive selection. The estimated selection coefficients are among the highest known in recent human evolution.

In other words: where people truly depended on milk as a staple resource, natural selection strongly favoured lactase persistence. Where milk was only one component of a mixed subsistence economy, it did not.

As Johannes Krause (Max Planck Institute for Evolutionary Anthropology) nicely puts it: “To me, it’s a nail in the coffin to the idea that it wasn’t milk, but something else, that has driven its frequency.”

This study elegantly reconciles archaeology, population genetics and evolutionary theory: lactase persistence is not a universal story of selection wherever milk is used, but a highly context-dependent one. Yet, when human survival really hinges on dairy, milk alone is more than enough to leave one of the strongest footprints of selection in our genome.

1. <https://pubmed.ncbi.nlm.nih.gov/22948027/>
2. <https://www.ncbi.nlm.nih.gov/pubmed/41278663>