Lactose Tolerance
From “Before the Dawn” by Nicholas Wade

Geneticists are still trying to define the exact genetic change that causes the lactase gene to stay active after weaning. The DNA sequence of the lactase gene itself is identical in both lactose tolerant and intolerant people. The difference must lie in some nearby region of DNA that controls the activation of the lactase gene, such as the two mutations recently discovered by Leena Peltonen* of the University of Helsinki.

What is certain is that the lactose tolerant Europeans have inherited unchanged from a common ancestor a huge block of DNA that includes the lactase gene, its neighboring gene, and much else (located on chromosome 2). The size of the block is a sign of recent evolutionary change. Big blocks of unchanged DNA are very rare because at each generation pairs of chromosomes swap sections of DNA so as to create individuals with novel combinations of genes. As is easy to envisage, the blocks of original DNA that a chromosome may start off with will get smaller and smaller at each generation as the swapping process whittles them down. So a large block of DNA shared by lots of people is a sign of recent selection. Large blocks are created when some must-have mutation occurs that is greatly favored by natural selection. Nature cannot pick out a specific mutation or gene; it can only favor individuals who have inherited the large block of DNA within which the advantageous gene occurs.

Besides indicating the presence of a gene under natural selection, a block of DNA can also be used to date the time the gene started to be selected, since the larger it is, the more recent the selection. Joel Hirschhorn** of the Harvard Medical School has found that the block containing the lactase gene in lactose tolerant Europeans extends for about 1 million DNA units. He and colleagues believe that this is a sign of strong positive selection, and that the block started to become widespread sometime between 2,000 and 21,000 years ago. This date fits with that of the Funnel Beaker culture.

Lactose tolerance occurs in a high percentage of many northern Europeans who live in the former region of the Funnel Beaker culture – in 100% of Dutch people, according to one survey, and 99% of Swedes. The condition also occurs in many other populations, though at generally much lower rates. In Africa, tribes who keep cattle, sheep or goats have higher rates of lactose tolerance than nonpastoralists. Lactose tolerance in some African groups includes as much as 25% of the population. It is presumably less common in these African groups than in northern Europeans because pastoralism got started later in Africa and natural selection has had less time to raise the frequency of the gene.

Lactose tolerance seems to have a different genetic basis in Africa because the DNA differences found by Peltonen and colleagues to be diagnostic of lactose tolerance in Europeans are largely absent from Africa.

The phenomenon of lactose tolerance draws attention to three aspects of human evolution. First, it confirms that evolution didn’t stop 50,000 years ago, when modern humans left Africa, as is often assumed, but has continued to reshape the human genome.

Second, it shows the human genome is likely to have responded independently in different populations to the same stimulus, a process known as convergent evolution. Lactose tolerance has arisen independently in northern Europeans and in several African populations. Many other human attributes that have evolved since the African diaspora may also have taken place independently in different populations, such as the probably cognitive advances discussed in Chapter 5.
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Third, the lactose tolerance phenomenon establishes that genes respond to cultural changes. This is not so surprising because culture is a major part of the human environment, and genomes are mechanisms for responding to the environment. But a feedback of culture on genes is rarely considered by social scientists, many of whom assume that human evolution ended for all practical purposes when cultural development began. The case of lactose tolerance shows that any long lasting human cultural behavior, such as drinking raw milk, can cause genetic changes if there is a way for the genome to respond to it.

Looking back on the years between 50,000 and 5,000 years ago, from the time of the ancestral human population to that of the Funnel Beaker people and their contemporaries, it is clear that wrenching changes in the human environment took place during this period, particularly in the social environment. Hunter-gatherers learned to settle down and cooperate in larger groups with people to whom they had no kin relationship. People who had been egalitarian and generalist joined hierarchical societies in which occupations were increasingly specialized. All these changes probably induced different behaviors, some of them maybe mediated through evolutionary changes to the human genome.

Human nature, in other words, has probably changed significantly.

*http://www.nature.com/ng/journal/v30/n2/pdf/ng826.pdf

Funnel Beaker Pottery
Figure 10.12  Distribution of adult lactose tolerance
People in areas with high lactose tolerance (such as Scandinavia) are likely to enjoy milk, cheese, and other dairy products throughout their lives. People in areas with low tolerance (such as much of Asia) do not ordinarily consume milk or dairy products as adults. (Source: Based on Flatz, 1987, and Rozin & Pelchat, 1988)