The New York Times
nytimes.com

December 10, 2006

Study Detects Recent Instance of Human Evolution

By NICHOLAS WADE

A surprisingly recent instance of human evolution has been detected among the peoples of East Africa. It is the ability to digest milk in adulthood, conferred by genetic changes that occurred as recently as 3,000 years ago, a team of geneticists has found.

The finding is a striking example of a cultural practice — the raising of dairy cattle — feeding back into the human genome. It also seems to be one of the first instances of convergent human evolution to be documented at the genetic level. Convergent evolution refers to two or more populations acquiring the same trait independently.

Throughout most of human history, the ability to digest lactose, the principal sugar of milk, has been switched off after weaning because there is no further need for the lactase enzyme that breaks the sugar apart. But when cattle were first domesticated 9,000 years ago and people later started to consume their milk as well as their meat, natural selection would have favored anyone with a mutation that kept the lactase gene switched on.

Such a mutation is known to have arisen among an early cattle-raising people, the Funnel Beaker culture, which flourished some 5,000 to 6,000 years ago in north-central Europe. People with a persistently active lactase gene have no problem digesting milk and are said to be lactose tolerant.

Almost all Dutch people and 99 percent of Swedes are lactose-tolerant, but the mutation becomes progressively less common in Europeans who live at increasing distance from the ancient Funnel Beaker region.

Geneticists wondered if the lactose tolerance mutation in Europeans, first identified in 2002, had arisen among pastoral peoples elsewhere. But it seemed to be largely absent from Africa, even though pastoral peoples there generally have some degree of tolerance.

A research team led by Sarah Tishkoff of the University of Maryland has now resolved much of the puzzle. After testing for lactose tolerance and genetic makeup among 43 ethnic groups of East Africa, she and her colleagues have found three new mutations, all independent of each other and of the European mutation, which keep the lactase gene permanently switched on.

The principal mutation, found among Nilo-Saharan-speaking ethnic groups of Kenya and Tanzania, arose 2,700 to 6,800 years ago, according to genetic estimates, Dr. Tishkoff's group is to report in the journal Nature Genetics on Monday. This fits well with archaeological evidence suggesting that pastoral peoples from the north reached northern Kenya about 4,500 years ago and southern Kenya and Tanzania 3,300 years ago.

Two other mutations were found, among the Beja people of northeastern Sudan and tribes of the same language family, Afro-Asiatic, in northern Kenya.

Genetic evidence shows that the mutations conferred an enormous selective advantage on their owners, enabling them to leave almost 10 times as many descendants as people without them. The mutations have created "one of the strongest genetic signatures of natural selection yet reported in humans," the researchers write.

The survival advantage was so powerful perhaps because those with the mutations not only gained extra energy from lactose but also, in drought conditions, would have benefited from the water in milk. People who were lactose-intolerant could have risked losing water from diarrhea, Dr. Tishkoff said.

Diane Gifford-Gonzalez, an archaeologist at the University of California, Santa Cruz, said the new findings were "very exciting" because they "showed the speed with which a genetic mutation can be favored under conditions of strong natural selection, demonstrating the possible rate of evolutionary change in humans."

The genetic data fitted in well, she said, with archaeological and linguistic evidence about the spread of pastoralism in Africa. The first clear evidence of cattle in Africa is from a site 8,000 years old in northwestern Sudan. Cattle there were domesticated independently from two other domestications, in the Near East and the Indus valley of India.

Both Nilo-Saharan speakers in Sudan and their Cushitic-speaking neighbors in the Red Sea hills probably domesticated cattle at the same time, since each has an independent vocabulary for cattle items, said Dr. Christopher Ehret, an expert on African languages and history at the University of California, Los Angeles. Descendants of each group moved southward and would have met again in Kenya, Dr. Ehret said.

Dr. Tishkoff detected lactose tolerance among both Cushitic speakers and Nilo-Saharan groups in Kenya. Cushitic is a branch of Afro-Asiatic, the language family that includes Arabic, Hebrew and ancient Egyptian.

Dr. Jonathan Pritchard, a statistical geneticist at the University of Chicago and the co-author of the new article, said that there were many signals of natural selection in the human genome, but that it was usually hard to know what was being selected for. In this case Dr. Tishkoff had clearly defined the driving force, he said.

The mutations Dr. Tishkoff detected are not in the lactase gene itself but a nearby region of the DNA that controls the activation of the gene. The finding that different ethnic groups in East Africa have different mutations is one instance of their varied evolutionary history and their exposure to many different selective pressures, Dr. Tishkoff said.

"There is a lot of genetic variation between groups in Africa, reflecting the different environments in which they live, from deserts to tropics, and their exposure to very different selective forces," she said.

People in different regions of the world have evolved independently since dispersing from the ancestral human population in northeast Africa 50,000 years ago, a process that has led to the emergence of different races. But much of this differentiation at the level of DNA may have led to the same physical result.

As Dr. Tishkoff has found in the case of lactose tolerance, evolution may use the different mutations available to it in each population to reach the same goal when each is subjected to the same selective pressure. "I think it's reasonable to assume this will be a more general paradigm," Dr. Pritchard said.