Jomon Genes

Using DNA, researchers probe the genetic origins of modern Japanese

By JOHN TRAVIS

Japan is generally considered one of the most modern, forward-looking countries in the world. Yet the Japanese also have an intense fascination with their islands' past, particularly the history and continuing influence of two peoples known as the Jomon and the Yayoi.

The Jomon, the original inhabitants of Japan, are thought to have migrated from the Asian mainland at a time when the two regions were physically connected. When sea levels began to rise about 12,000 years ago, the Japanese archipelago became separate from continental Asia, and the Jomon were left to spread across the islands. Hunters, fishers, and foragers, the Jomon were also the world's first known potters. Indeed, their name—Japanese for cord marks—stems from the ropelike impressions found in their clay pottery.

Scholars agree that the Jomon period of Japan's history ran from at least 10,000 years ago to about 250 B.C. At that point, the Yayoi, apparently traveling in ships from the Korean peninsula, arrived at the islands. The Yayoi culture, marked by weaving, metalworking, and, most important, the farming of rice, soon supplanted that of the Jomon.

During the last century, anthropologists have fiercely debated whether the Jomon or the Yayoi were the true ancestors of the modern Japanese. For most of this debate's history, scientists have addressed the issue by comparing Asian languages, analyzing archaeological ruins, and measuring dental or other skeletal remains.

Over the last few years, another kind of evidence has begun to have an impact on the debate. By studying the genes of modern Japanese and of other Asians, and even the ancient DNA in the fossilized bones of the Jomon and the Yayoi, investigators hope to put together a genetic history of Japan. Researchers have recently examined, for example, the Y chromosomes of people throughout Asia.

In conjunction with other historical evidence, the new work suggests that the Jomon did not originate in Southeast Asia, as one long-standing theory has it, but farther north. The research also sheds light on the extent to which the Jomon and the Yayoi have influenced the genetic makeup of modern Japanese.

"Our data clearly show that both Yayoi and Jomon genes have made a contribution to the contemporary gene pool," says Michael F. Hammer, who presented the Y chromosome research in October 1996 at the American Society of Human Genetics meeting in San Francisco.

Genetic studies suggest that Japan's original inhabitants, the Jomon, mixed with a later culture, the Yayoi. The Jomon's closest descendants today inhabit Japan's northern and southern islands.

Three theories have dominated the debate about the origin of the modern Japanese people. The replacement theory argues that the invading Yayoi wiped out the Jomon both culturally and genetically. Proponents of this option contend that the only remaining descendants of the Jomon are the Ainu in Hokkaido, the northernmost of the Japanese islands, and some inhabitants of Japan's southernmost islands. Among the physical features that make the Ainu distinct from most Japanese are lighter skin and more body hair.

"The Jomon are the obvious ancestors of the Ainu but not of modern Japanese," says C. Loring Brace, an anthropologist at the University in Michigan in Ann Arbor.

The transformation hypothesis holds that the Yayoi culture did supplant the Jomon culture but that the Yayoi did not come to Japan in large enough numbers to influence significantly the Jomon gene pool.

"Genetically, there's not much difference between the Jomon people and the current Japanese," asserts Masatoshi Nei, a population geneticist at Pennsylvania State University in State College and one of the strongest supporters of the transformation model.

The final theory offers a compromise. Usually referred to as the hybridization, or dual structure, model, it suggests that both the Yayoi and the Jomon have contributed significantly to the genes of most living Japanese.

Hammer, a researcher at the University of Arizona in Tucson, and Satoshi Horai, who works at the National Institute of Genetics in Mishima, Japan, addressed these theories with their study of the Y chromosome. In particular, the pair focused on an addition to the chromosome, the Y *Alu* polymorphic element, or YAP. *Alu* sequences are short stretches of DNA with the unusual ability to copy themselves and jump from one chromosome to another. The typical human genome holds an estimated 1 million *Alu* sequences, says Hammer.

YAP is an *Alu* sequence that hopped to a specific site on the Y chromosome at a relatively recent moment in human evolution. The addition of YAP seems to have occurred so recently that not every man in the world has it. "You're either a plus or a minus," Hammer explains.

The researchers began to realize that YAP might prove useful when an initial study of Asian populations revealed that only men from Japan seemed to harbor the genetic marker. In Taiwan and Korea, for example, not a single man was found to possess YAP, Hammer and Horai reported in the AMERICAN JOURNAL OF

HUMAN GENETICS. Furthermore, the pair found that YAP-positive chromosomes appeared with much greater frequency in the southern islands of Japan than in the country's main islands.

From that evidence, Hammer and Horai hypothesized that the YAP element was originally carried to Japan by the Jomon and that the Yayoi, who came from the region that now makes up North and South Korea, lacked the marker. More recent research has strengthened this theory.

Working with several colleagues, the two researchers mapped the distribution of YAP-positive chromosomes throughout Japan. While men living in central Japan rarely carry YAP, the Ainu and inhabitants of the southern islands, the two populations apparently least influenced by the Yayoi, frequently do.

Hammer and his colleagues are also studying a second Y chromosome marker that may serve as a sign of the Yayoi migration. This marker is common in Koreans and appears most frequently in the central islands of Japan, says Hammer.

Together, contends Hammer, the two markers tell a story of an initial Yayoi migration into central Japan and a subsequent spread of the people toward the north and south. Since both Y chromosome markers are still found in varying degrees throughout Japan, it appears that the genes of the Jomon and Yayoi peoples did intermingle significantly. "Our data support the hybridization theory," says Hammer.

The research on YAP has also addressed another controversial question: Where did the Jomon come from? Some researchers have long held the idea that the Jomon originated in southeast Asia and spread to Japan about 12,000 years ago. Analyses of dental remains, shared aspects of language, and even some genetic studies have offered support for this scenario.

Several years ago, Nei offered an alternative. Working from his own analysis of more than a dozen genetic markers on a variety of chromosomes and from archaeological data showing habitation of Japan dating back 30,000 years, Nei argued that the Jomon actually came from northeastern Asia and settled in Japan far earlier than supporters of the Southeast Asia theory had proposed.

While the YAP data do not appear to support the transformation theory favored by Nei, they may bolster Nei's vision of the Jomon's origin, says Hammer. He and his colleagues surveyed more than 1,000 men from 20 populations in Southeast Asia. "We didn't find a single example of a YAP-positive chromosome," says Hammer.

In contrast, a survey of 700 men from 13 northern Asian populations did reveal areas with YAP-positive chromosomes. About 3 percent of men from southwestern Siberia and Mongolia have the marker. The most spectacular finding was that more than 50 percent of men from Tibet harbor the YAP element.

While Tibet is part of central Asia, Nei suggests that the YAP data support his theory that the Jomon originated in the northeast. Other researchers have concluded that the Tibetans arrived in their current homeland only several thousand years ago, after a migration from northeast Asia, Nei points out.

The new Y chromosome research by Hammer and Horai is unlikely to settle the century-old debate about the origin of the Jomon and their genetic contribution to modern Japanese, caution researchers following the issue.

"You may get a different story when you look at the Y chromosome than when you look at mitochondrial DNA or when you look at other nuclear genes," warns Brace, who adds that any genetic data must also be reconciled with traditional archaeological evidence. "They all have to mesh to tell a complete story."

Unraveling that story continues to hold a high priority in Japan. Over the next 4 years, the country's Ministry of Education intends to fund a large, multidisciplinary research effort to examine the origins of the Japanese and their culture. □