
Chapter 9

Genetic Relationships of Human Populations in and around the Japanese Archipelago

Saitou Naruya

Dedicated to the memory of the late Hanihara Kazuro

Genetic and linguistic studies on the phylogenetic relationships of populations in and around the Japanese Archipelago are presented. The genetic data suggest a weak affinity between Ainu and Okinawan, compatible with the Dual Structure hypothesis of the late Hanihara Kazuro. Linguistically, Okinawan is a Japanese dialect, and the Ainu language is distant from Japanese. However, some possibility of similarity between the Ainu and Okinawa languages is suggested through phylogenetic network analysis.

The Japanese Archipelago consists of four major islands (Hokkaido, Honshu, Shikoku, and Kyushu) and many surrounding small islands (Fig. 9.1). Owing to its vicinity to the Eurasian Continent, frequent waves of migration from the continent to the islands took place during at least the last 50,000 years. There are six major migration routes to the Japanese Archipelago (Fig. 9.1). Route 1, the most plausible pathway, is through the Korean Peninsula, and the next possible one, route 2, is via Sakhalin Island. The northernmost route 3 is via the Kamchatkan Peninsula and the Chishima (Kurile) Archipelago, while the southernmost route 4 is via Taiwan and the Ryukyu Archipelago. In Japan route 4 is known as the 'Sea Road' (Yanagita 1961). There are two other routes, but both of them need relatively long crossings of either the East China Sea (route 5) or the Sea of Japan (route 6), and probably became important only recently, perhaps within the past 3000 years. There is another possible human migration, from the Pacific Ocean, i.e. visitors from Polynesia, but the relative contribution to the genetic constitution of the people in the Japanese Archipelago is small.

It may be pertinent to divide the population of the Japanese Archipelago into three groups in terms of their cultural and historical perspectives; the Ainu, the mainland Japanese, and the Okinawans. The Ainu people, who currently live mainly in Hokkaido, also lived in Sakhalin and the Chishima Archipelago until

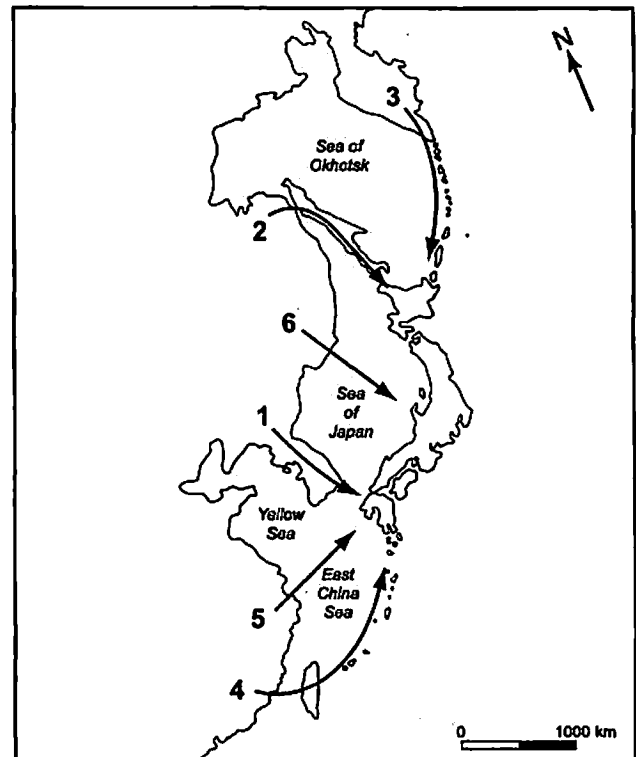


Figure 9.1. Six possible routes of migration to the Japanese Archipelago. (Drawing: D. Kemp from Saitou 2005.)

Table 9.1. Cultural periods of the central part of the Japanese Archipelago. (From Saitou 2005.)

Period	Time
Palaeolithic	~13,000 BP
Jomon	13,000–3000 BP
Yayoi	3000–1800 BP
Kofun-Historical	1800 BP–Present

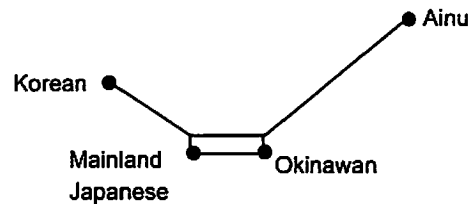
recently. The Ainu population was about 23,000 in 1993 (Ainu Museum web; www.ainu-museum.or.jp/), and Hokkaido is now mostly populated by immigrants from the mainland (Honshu and the other two major islands). Mainland Japanese form the majority of current 'Japanese', and their population size was approximately 130 million in 2005. Okinawans are the majority in the Ryukyu Archipelago, but there are many migrants and their descendants from mainland Japan. Although there are now several ancient (more than 15,000 BP) human remains found from Okinawa, the real start of Okinawan prehistory was about 6000 BP (Asato & Doi 1999).

Table 9.1 shows the historical framework of the central part of the Japanese Archipelago. Archaeological details of these prehistoric periods are reviewed in Imamura (1996). Hanihara (1991) proposed a 'Dual Structure' hypothesis to explain the current geographical distribution pattern of the Japanese population. This hypothesis consists of two parts: 1) that the Upper Palaeolithic population of Japan that gave rise to the Jomon people was of southeast Asian origin; and 2) that the modern Ainu and Okinawa populations are direct descendants of the Jomon people, while mainland Japanese are mainly derived from migrants from the northeast Asian continent after the Yayoi period.

Genetic studies

Omoto & Saitou (1997) studied the genetic relationship of three human populations (Ainu, mainland Japanese, and Okinawan) in Japan and Korean using 25 classic markers. When they used the neighbour-joining method (Saitou & Nei 1987), Ainu and Okinawans became neighbours, as did mainland Japanese with Koreans. Bootstrap probabilities for clustering between these two populations were 85% and 74% for the DA distance and the Dst distance, respectively, somewhat lower than statistically significance levels. However, those bootstrap probabilities were much higher than the random expectation (33%). Omoto & Saitou (1997) therefore provided partial support for Hanihara's (1991) Dual Structure hypothesis for explaining the genetic structure of the Japanese.

(A) Genetic Network



(B) Language Network

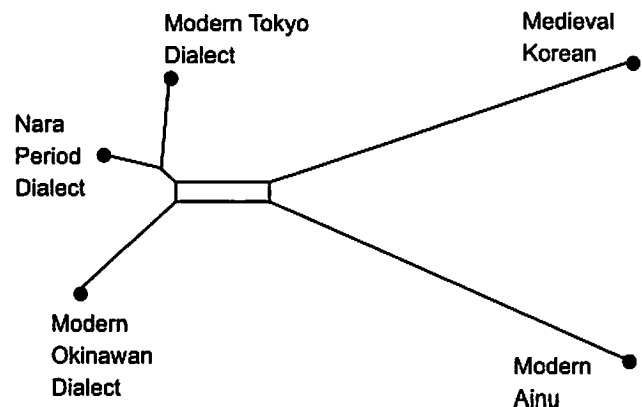


Figure 9.2. Phylogenetic networks for (A) genetic data and for (B) language data. ((A) is based on Omoto & Saitou (1997), and (B) is from Saitou (2005).)

Figure 9.2A shows a split decomposition network (Bandelt & Dress 1992) based on the DA distances given in Omoto & Saitou (1997). The split ((Ainu, Okinawan)–(Korean, mainland Japanese)) is longest, followed by ((Okinawan, mainland Japanese)–(Ainu, Korean)). The remaining possible split ((Ainu, mainland Japanese)–(Okinawan, Korean)) does not exist in this network. This network suggests that Ainu and Okinawan share some genetic similarity, even though Ainu is quite distantly related from the three other populations. The similarity between mainland Japanese and Okinawan, shown by the second split, suggests the existence of gene flow between these two populations. The genetic similarity between Ainu and Okinawan supports Hanihara's (1991) dual structure hypothesis on the origin of the Japanese at least partially.

A similar relationship was also observed for the Y chromosome (Hammer & Horai 1995), HLA (Bannai *et al.* 2000), and for mtDNA (Tajima *et al.* 2003).

Tajima and colleagues (2003) determined mtDNA sequences for Ainu and some other human populations surrounding Japan, and they found that haplo-

types 2 and 4 found in the Ainu were also found in the Nivkhi and Kamchatkan populations, respectively. This clearly indicates a genetic influence of North-eastern Asian populations on the Ainu.

Tanaka and colleagues (2004) determined complete mitochondrial DNA sequences for 672 Japanese individuals and compared them with many other Asian sequences. They also found a clear genetic similarity between mainland Japanese and Asian continental populations such as Korean and Han Chinese.

Li and colleagues (2005) recently examined more than 100 STR loci for five Han Chinese populations and two Japanese populations (mainland and Okinawa). Two Japanese populations were tightly clustered with 100% bootstrap probabilities, but interestingly, this Japanese cluster was more similar to southern Han Chinese populations than to northern ones. This pattern is somewhat different from the phylogenetic relationship of Chinese and surrounding Asian populations using HLA data (Saitou *et al.* 1992), where both Japanese and Korean were genetically much closer to northern Han Chinese. However, when Alu insertion/deletion polymorphism was used for the same Han Chinese samples and Japanese, the northern Han were somewhat closer to the Japanese (Ishibashi and Saitou, unpublished). This discrepancy is worth examining in future studies.

Linguistic studies

The origin of the Japanese language has long been debated. Arai Hakuseki, a high-ranking Tokugawa Shogun government official in the eighteenth century, suggested that Japanese was similar to the Korean language, and since then many people have proposed an affinity of these two languages. Some linguists considered these two languages as well as the Ainu language as belonging to the Altaic language family (see Vovin & Osada 2003). The Okinawan language is considered to be a dialect of Japanese because of their close relationship, while the phylogenetic language relationship between Ainu and Japanese is not clear; some linguists such as Kindaichi Kyosuke and Chiri Mashio concluded that these two languages were not related, while others such as Murayama Shichiro and Hattori Shiro accepted a weak similarity of these two languages, though indirectly (Saitou 2005). It seems that the majority of linguists do not currently conduct numerical and statistical analyses in studying the phylogenetic relationship of languages.

Yasumoto & Honda (1978) compared lists of 100 and 200 basic words of many languages around the Japanese Archipelago, and proposed the existence of a common ancestral language for the contemporary

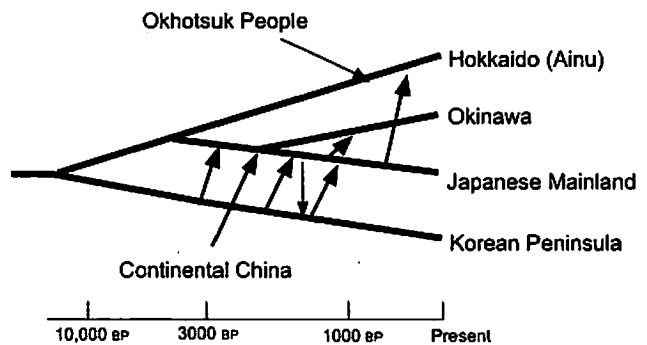


Figure 9.3. Scenario for temporal changes of human populations in the Japanese Archipelago and the Korean Peninsula. (From Saitou 2005.)

Japanese, Ainu, and Korean languages based on their results. Saitou (2005) constructed a phylogenetic network (Fig. 9.2B) for these three languages (three dialects for Japanese, Ainu, and medieval Korean) using data collected by Yasumoto & Honda (1978). As expected, the three Japanese dialects clearly formed one cluster, in particular, the Nara period (eighth century AD) dialect was closest to contemporary Japanese, followed by the contemporary dialect of Okinawa's Shuri area. Interestingly, there were three words that showed similarity between the Okinawan and Ainu languages.

Although the genetic and linguistic relationships of human populations in and around the Japanese Archipelago are not consistent, this difference itself may tell something about the history of human populations in this region. Figure 9.3 summarizes one plausible scenario of temporal changes of human populations in the Japanese Archipelago. I hope this can explain both genetic and linguistic patterns shown in Figure 9.2.

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References

- Asato, S. & N. Doi, 1999. *Okinawajinwa Dokokara Kitaka* [Where did the Okinawan People Originate?]. Naha: Border. [In Japanese.]
- Bandelt, H.-J. & A.W. Dress, 1992. Split decomposition: a new and useful approach to phylogenetic analysis of

- distance data. *Molecular Phylogenetics and Evolution* 1, 242–52.
- Bannai, M., J. Ohashi, S. Harihara, *et al.*, 2000. Analysis of HLA genes and haplotypes in Ainu (from Hokkaido, northern Japan) supports the premise that they descent from Upper Paleolithic populations of East Asia. *Tissue Antigens* 55, 128–39.
- Hammer, M.F. & S. Horai, 1995. Y chromosomal DNA variation and the peopling of Japan. *American Journal of Human Genetics* 56, 951–62.
- Hanihara, K., 1991. Dual structure model for the population history of the Japanese. *Japan Review* 2, 1–33.
- Imamura, K., 1996. *Prehistoric Japan*. Honolulu (HI): University of Hawaii Press.
- Li, S.-L., T. Yamamoto, T. Yoshimoto, *et al.*, 2005. Phylogenetic relationship of the populations within and around Japan using 105 short tandem repeat polymorphic loci. *Human Genetics* 118(6), 695–707.
- Omoto, K. & N. Saitou, 1997. Genetic origins of the Japanese: a partial support for the ‘dual structure hypothesis’. *American Journal of Physical Anthropology* 102(4), 437–46.
- Saitou, N., 2005. *DNA Kara Mita Nihonjin* [Japanese Viewed from DNA]. Tokyo: Chikuma Shobo. [In Japanese.]
- Saitou, N. & M. Nei, 1987. The neighbor-joining method: a new method for reconstructing phylogenetic trees. *Molecular Biology and Evolution* 4(4), 406–25.
- Saitou, N., K. Tokunaga & K. Omoto, 1992. Genetic affinities of human populations, in *Isolation, Migration, and Health: 33rd Symposium Volume of the Society for the Study of Human Biology*, eds. D.F. Roberts, N. Fujiki & K. Torizuka. New York (NY): Cambridge University Press, 118–29.
- Tajima, A., M. Hayami, K. Tokunaga, *et al.*, 2003. Genetic origins of the Ainu inferred from combined DNA analyses of maternal and paternal lineages. *Journal of Human Genetics* 49, 187–93.
- Tanaka, M., V.M. Cabrera, A.M. Gonzalez, *et al.*, 2004. Mitochondrial genome variation in eastern Asia and the peopling of Japan. *Genome Research* 14, 1832–50.
- Vovin, A. & T. Osada (eds.), 2003. *Perspectives on the Origin of the Japanese Language*. (Nichibunken Japanese Studies Series 31.) Kyoto: International Research Center for Japanese Studies. [In English and Japanese.]
- Yanagita, K., 1961. *Kaijo no Michi* [Sea Road]. Tokyo: Chikuma Shobo. [In Japanese.]
- Yasumoto, B. & M. Honda, 1978. *Nihongo No Tanjo* [Emergence of the Japanese Language]. Tokyo: Taishukan Shoten. [In Japanese.]