

COMMENTARY

A commentary on the history of human populations in the Japanese Archipelago inferred from genome-wide SNP data with a special reference to the Ainu and the Ryukyuan populations

Jun Gojobori

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The first population to occupy the Japan Archipelago migrated there more than 30 000 years ago and established the Jomon culture around 12 000 years ago.^{1,2} The second wave of migrants, which possessed advanced culture such as wet rice agriculture, weaving and metalworking, migrated to Japan around 2300 years ago and they gave rise to the Yayoi culture.² Hanihara³ proposed the ‘dual structure’ hypothesis, which explains the origin of the current Japanese population by proposing an admixture of these two major migrant populations, rather than a transition of the Jomon into the Yayoi. Hanihara’s hypothesis is supported by the morphological, cultural and genetic similarity between Ryuku and Ainu peoples, whose populations are separated by considerable distance, with the Ryuku residing in Okinawa and Ainu in Hokkaido. Assuming the Ryuku and Ainu populations have less genetic influence from Yayoi migrants compared with Mainland Japanese, the similarity between them can be explained by the higher contribution from the Jomon people they have in common. The ‘dual structure’ hypothesis received further support from genetical studies,^{4,5} but its simple dichotomy—Jomon people as hunter-gatherers and Yayoi people as farmers—has been questioned.⁶

Using high-throughput SNP genotyping, a study of genome-wide genetic diversity in the current Japanese population was conducted.⁷

This new study by the Japanese Archipelago Human Population Genetics Consortium, which appeared in the *Journal of Human Genetics*, is a solid addition to the body of literature pertaining to the study of the Japanese people.⁸ This study provides further insights into the origin of the Japanese people by including hundreds of thousands of SNP data from Ainu, which were not included in the earlier studies.⁷ Two of the consortium member visited Biratori town and explained this study to the representatives of the Ainu people living in that area. Together with the samples of Mainland Japanese, Ryuku and the other population from the rest of the world, they conducted primary components analysis (PCA), frappe analysis and phylogeny analysis including network studies to reveal

the relationship between Mainland Japanese, Ainu and Ryuku.

One of the findings of the study was the close relationship between the Ainu and Ryuku (Figure 1). In a phylogenetic tree, the Ainu and Ryuku cluster together (100% bootstrap support) with the Mainland Japanese forming a sister group to them. This phylogenetic relationship supports the ‘dual structure’ hypothesis³ where previous studies could only provide partial support.⁴ The authors also observed recent admixture of Ainu with Mainland Japanese, but they suggest the possibility of additional admixture with another population. The authors concluded that the Ainu experienced two types of admixtures but the source population other than Mainland Japanese could not be identified. This conclusion was

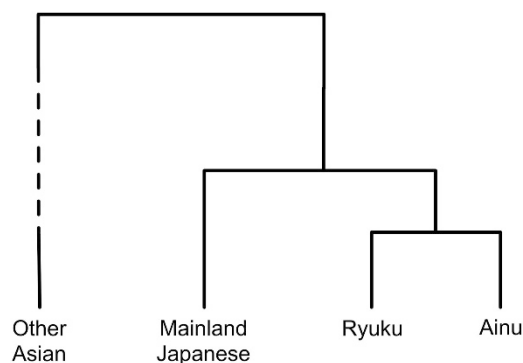


Figure 1 A schematic illustration of relationship among three populations in Japan, Mainland Japanese, Ryuku and Ainu. The close relationship between Ryuku and Ainu is supported by Japanese Archipelago Human Population Genetics Consortium.⁸ This result supports the ‘dual structure’ hypothesis proposed by Hanihara.³

Dr J Gojobori is at The School of Advanced Sciences, Graduate University for Advanced Studies, Kanagawa, Japan.
E-mail: gojobori_jun@soken.ac.jp

drawn by the PCA plots and frappe analysis. The study also showed that the Ainu have higher genetic heterogeneity compared with Mainland Japanese or Ryuku. However, the authors suggest that this heterogeneity is possibly due to degradation of Ainu DNA samples since their collection nearly 30 years ago.

Key questions raised by this study are 'What population is the source of admixture in the Ainu other than Mainland Japanese?' and 'Do the Ainu truly have higher genetic heterogeneity?' Collecting more population samples from various Asian regions will be helpful to answer to the first question, whereas collecting fresh Ainu DNA samples, if possible, will address the second question. Ancient DNA sample from both the Jomon and Yayoi ages may further our understanding of the origin of the Japanese people as it might be possible to observe the chronological changes of genetic variation of the occupants of Japan archipelago. A study by Skuglund *et al.*⁹ that addresses the spread of agriculture in Europe is a good example of a study that uses ancient DNA.

As mentioned in the study by Skuglund *et al.*,⁹ whether the transition from hunter-gatherer to agriculture was done in a peaceful way or not is subject to debate.⁹ A key question is whether farmers drove hunters away or hunter-gatherers themselves acquired agriculture and became farmers. This question relates to the transition of Jomon culture to Yayoi culture and the origin of agriculture in Japan. That is, whether the Jomon people became the Yayoi people or the current Japanese population is an admixture of both.¹⁰ Insights into the origin of Japanese agriculture can be gained by understanding the genetic influence of the relatively advanced Yayoi people on the more ancient Jomon people. The peopling of the Japan archipelago might be more complicated than previously thought⁶ and the Ainu may not have been simple hunter-gatherers.¹⁰ This study by the Japanese Archipelago Human Population Genetics Consortium⁸ is an important first step toward understanding the genetic variation among the Ainu people and will contribute to our understanding of the peopling of the Japan archipelago.

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