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ORIGINAL ARTICLE

A partial nuclear genome of the Jomons who lived 3000 years ago in Fukushima, Japan

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The Jomon period of the Japanese Archipelago, characterized by cord-marked 'jomon' potteries, has yielded abundant human skeletal remains. However, the genetic origins of the Jomon people and their relationships with modern populations have not been clarified. We determined a total of 115 million base pair nuclear genome sequences from two Jomon individuals (male and female each) from the Sanganji Shell Mound (dated 3000 years before present) with the Jomon-characteristic mitochondrial DNA haplogroup N9b, and compared these nuclear genome sequences with those of worldwide populations. We found that the Jomon population lineage is best considered to have diverged before diversification of present-day East Eurasian populations, with no evidence of gene flow events between the Jomon and other continental populations. This suggests that the Sanganji Jomon people descended from an early phase of population dispersals in East Asia. We also estimated that the modern mainland Japanese inherited <20% of Jomon peoples' genomes. Our findings, based on the first analysis of Jomon nuclear genome sequence data, firmly demonstrate that the modern mainland Japanese resulted from genetic admixture of the indigenous Jomon people and later migrants.

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INTRODUCTION

Genome-wide single-nucleotide polymorphism (SNP) data analyses[1,2] support the dual-structure model[3,4] for the formation of modern Japanese populations, in which indigenous Jomon people admixed with later migrants who brought rice agriculture. The Jomon culture geographically ranged from Hokkaido to the Okinawa islands, and the Jomon people inhabited the Japanese Archipelago from ~16000 to 2500 years before present (YBP).[5,6] The origins and phylogenetic relationships of the Jomon people, however, are still elusive.

Ancient DNA sequences of the Jomon people provide direct evidence of their genetic characteristics. Mitochondrial DNA (mtDNA) sequences of the Jomon people and their haplotypes have been determined for many individuals.[7–12] Haplogroup N9b, whose frequency is generally low in modern East Eurasians,[13,14] was found to be quite frequent in the mtDNA of the Jomons.[9–11] This suggests a long-term isolation of the Jomons from continental populations. However, inferring human population history only from mtDNA data are insufficient because of their limited genetic information. Thanks to new technologies, it is now possible to analyze nuclear genome sequences of ancient human remains[15–23] and those of modern human individuals.[24] We therefore determined the nuclear genome sequences of two Jomon individuals and compared them with available data so as to infer the origin of modern Japanese.

MATERIALS AND METHODS

DNA extraction, library preparation, sequencing and sequence mapping

We extracted DNA from the teeth of Jomon individuals kept at the University Museum, the University of Tokyo. These samples were originally excavated at the Sanganji Shell Mound in the 1950s.[25] This shell mound is located in the northern part of Fukushima Prefecture, Tohoku region, Japan (Supplementary Figure 1). The mtDNA haplotypes of four Sanganji Jomon individuals were previously genotyped.[11] We designate two DNA extracts of the Sanganji 131421-3 individual, one extracted by Kanzawa-Kiriyama *et al.*,[11] and the other newly extracted for this study, as A1 and A2, respectively. The DNA extract from sample 131464 is referred to as B. The new DNA extract (A2) was prepared using Adachi *et al.*'s[12] protocol with some modifications (Supplementary Methods S1). Three DNA libraries for A1, A2 and B were prepared from the DNA solution extracted from their molars (Supplementary Methods S1). The GAIIx platform was used for library A1, and 120-bp paired-end sequence reads were generated following the manufacturer's protocol.

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