

## ORIGINAL ARTICLE

# Unique characteristics of the Ainu population in Northern Japan

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Various genetic data (classic markers, mitochondrial DNAs, Y chromosomes and genome-wide single-nucleotide polymorphisms (SNPs)) have confirmed the coexistence of three major human populations on the Japanese Archipelago: Ainu in Hokkaido, Ryukyuan in the Southern Islands and Mainland Japanese. We compared genome-wide SNP data of the Ainu, Ryukyuan and Mainland Japanese, and found the following results: (1) the Ainu are genetically different from Mainland Japanese living in Tohoku, the northern part of Honshu Island; (2) using Ainu as descendants of the Jomon people and continental Asians (Han Chinese, Koreans) as descendants of Yayoi people, the proportion of Jomon genetic component in Mainland Japanese was ~18% and ~28% in Ryukyuan; (3) the time since admixture for Mainland Japanese ranged from 55 to 58 generations ago, and 43 to 44 generations ago for the Ryukyuan, depending on the number of Ainu individuals with varying rates of recent admixture with Mainland Japanese; (4) estimated haplotypes of some Ainu individuals suggested relatively long-term admixture with Mainland Japanese; and (5) highly differentiated genomic regions between Ainu and Mainland Japanese included *EDAR* and *COL7A1* gene regions, which were shown to influence macroscopic phenotypes. These results clearly demonstrate the unique status of the Ainu and Ryukyuan people within East Asia.

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## INTRODUCTION

The Japanese Archipelago consists of four major islands (Hokkaido, Honshu, Shikoku and Kyushu) and many other small islands that can be grouped into nine regions (Supplementary Figure 1). Frequent waves of human migrations from the Eurasian continent to the archipelago took place from at least 30 000 years ago (YBP).<sup>1</sup> There were various migration routes to the archipelago.<sup>2</sup> These migrations have shaped the human population structure in the Japanese Archipelago, where there are currently three main populations: the Ainu who mainly live in Hokkaido at the northernmost island of the Archipelago; the Ryukyuan who mainly live in the Ryukyu Islands at the southern part; and the Mainland Japanese whose population size is the largest and who live in all major four islands and small islands.

From an archeological perspective, the prehistory of the Japanese Archipelago can be divided into the Paleolithic period (older than 16 000 YBP), the Jomon period (16 000–3000 YBP) and the Yayoi period (3000–1700 YBP).<sup>1</sup> The currently accepted model regarding the origin of Japanese populations is the dual-structure model,<sup>3</sup> whereby the current Japanese population is the result of admixture between the early migrants (Jomon people) and later migrants (Yayoi people) and that the Ainu and the Ryukyuan are thought to retain more Jomon components than the Mainland Japanese. Subsequent studies using

mitochondrial DNA and several autosomal markers have been in general agreement with the dual-structure model, showing the admixed nature of Mainland Japanese<sup>4,5</sup> and demonstrating close affinities between the Ainu and Ryukyuan populations.<sup>6,7</sup>

The Japanese Archipelago Human Population Genetics Consortium<sup>8</sup> produced ~900 000 genome-wide single-nucleotide polymorphism (SNP) data in the Ainu and the Ryukyuan, and through principal component analysis (PCA), and phylogenetic tree construction, demonstrated a clear genetic similarity between these two groups despite their current geographical locations at the opposite poles of the Japanese archipelago. Analysis of individual ancestry proportions and phylogenetic analysis of the Mainland Japanese also show that they carry both Ainu-Ryukyuan and continental Asian genetic components. A recent study that used a model-based approach was also in favor of the dual-structure model.<sup>9</sup>

Although previous studies generally support the dual-structure model, some details regarding the amount of genetic contributions from the ancestral populations were not really well defined. We also wanted to identify what kind of factors that contributed to the genetic uniqueness that was previously observed in the Ainu.<sup>8</sup> Therefore, the aims of this study are to perform a test for admixture and to clarify the timing and admixture proportions in the Japanese populations.

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