

ORIGINAL ARTICLE

# 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

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The Japanese Archipelago stretches over 4000 km from north to south, and is the homeland of the three human populations; the Ainu, the Mainland Japanese and the Ryukyuan. The archeological evidence of human residence on this Archipelago goes back to > 30 000 years, and various migration routes and root populations have been proposed. Here, we determined close to one million single-nucleotide polymorphisms (SNPs) for the Ainu and the Ryukyuan, and compared these with existing data sets. This is the first report of these genome-wide SNP data. Major findings are: (1) Recent admixture with the Mainland Japanese was observed for more than one third of the Ainu individuals from principal component analysis and frappe analyses; (2) The Ainu population seems to have experienced admixture with another population, and a combination of two types of admixtures is the unique characteristics of this population; (3) The Ainu and the Ryukyuan are tightly clustered with 100% bootstrap probability followed by the Mainland Japanese in the phylogenetic trees of East Eurasian populations. These results clearly support the dual structure model on the Japanese Archipelago populations, though the origins of the Jomon and the Yayoi people still remain to be solved.

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## INTRODUCTION

The origins of the people living in the Japanese Archipelago have been studied for a long time (for review, see Suzuki<sup>1</sup> and Saitou<sup>2</sup>). The standard theory based on craniofacial data is the dual structure

model proposed by Hanihara.<sup>3</sup> According to this model, the first migrants to the Japanese Archipelago came from somewhere in Southeast Asia in the Upper Paleolithic age, who were ancestors of the Jomon people. The second wave of migration took place later in

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