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Mitochondrial DNA evidence supports northeast Indian origin of the aboriginal Andamanese in the Late Paleolithic

Hua-Wei Wang [a], Bikash Mitra [a,b], Tapas Kumar Chaudhuri [b], Malliya gounder Palanichamy [a], Qing-Peng Kong [c,d,*], Ya-Ping Zhang [a,c,d,*]

[a] *Laboratory for Conservation and Utilization of Bio-resource, Yunnan University, Kunming 650091, Yunnan Province, China*
[b] *Cellular Immunology Laboratory Department of Zoology University of North Bengal, Siliguri, West Bengal 734013, India*
[c] *State Key Laboratory of Genetic Resources and Evolution, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming 650223, Yunnan Province, China*
[d] *KIZ/CUHK Joint Laboratory of Bioresources and Molecular Research in Common Diseases, Kunming 650223, Yunnan Province, China*

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Abstract

In view of the geographically closest location to Andaman archipelago, Myanmar was suggested to be the origin place of aboriginal Andamanese. However, for lacking any genetic information from this region, which has prevented to resolve the dispute on whether the aboriginal Andamanese were originated from mainland India or Myanmar. To solve this question and better understand the origin of the aboriginal Andamanese, we screened for haplogroups M31 (from which Andaman-specific lineage M31a1 branched off) and M32 among 846 mitochondrial DNAs (mtDNAs) sampled across Myanmar. As a result, two Myanmar individuals belonging to haplogroup M31 were identified, and completely sequencing the entire mtDNA genomes of both samples testified that the two M31 individuals observed in Myanmar were probably attributed to the recent gene flow from northeast India populations. Since no root lineages of haplogroup M31 or M32 were observed in Myanmar, it is unlikely that Myanmar may serve as the source place of the aboriginal Andamanese. To get further insight into the origin of this unique population, the detailed phylogenetic and phylogeographic analyses were performed by including additional 7 new entire mtDNA genomes and 113 M31 mtDNAs pinpointed from South Asian populations, and the results suggested that Andaman-specific M31a1 could in fact trace its origin to northeast India. Time estimation results further indicated that the Andaman archipelago was likely settled by modern humans from northeast India *via* the land-bridge which connected the Andaman archipelago and Myanmar around the Last Glacial Maximum (LGM), a scenario in well agreement with the evidence from linguistic and palaeoclimate studies.

Keywords: mtDNA; Myanmar; Aboriginal Andamanese; Northeast India; Origin

1. Introduction

Due to their "Negritos" characters, the aboriginal Andamanese once were believed to have close affinity with African. However, according to the genetic evidence from Y chromosome and mtDNA, the aboriginal Andamanese showed closer affiliation to Asian than to African (Thangaraj et al., 2003); whereas the genetic work based on the museum samples also illustrated the close genetic affinity between the aboriginal Andamanese and South Asian populations (Endicott et al., 2003). This unique population recently was even suggested to be derived directly from the initial modern humans when they migrated along Asian coast (Thangaraj et al., 2005). However, this notion was questioned largely due to the observation of a sister clade of the claimed Andamanese-specific haplogroup, M31, in Indian sub-continent (Palanichamy et al., 2006). The latter finding instead suggested that the aboriginal Andamanese likely traced their origin to an already differentiated human population residing

* Corresponding authors. Tel: +86 871 503 2804, fax: +86 871 503 2804 (Y-P Zhang); Tel: +86 0871 519 7967, fax: +86 0871 519 7967 (Q-P Kong).
E-mail addresses: kongqp@yahoo.com.cn (Q-P Kong), zhangyp1@263.net.cn (Y-P Zhang).

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somewhere in India (Palanichamy et al., 2006). Thereafter, more and more M31 lineages were observed in South Asian populations (Endicott et al., 2006; Reddy et al., 2007; Barik et al., 2008; Fornarino et al., 2009), which all suggested northeast India likely being the origin place of haplogroup M31. Therefore, it seems that northeast India may represent the source place of the ancestors of aboriginal Andamanese. However, all the abovementioned studies on haplogroup M31 mainly focused on populations sampled from India and Nepal; so far there is no any genetic information from Myanmar reported elsewhere. In consideration of the fact that Myanmar is the closest region to the Andaman archipelago in geographic, without genetic data from Myanmar, it is impossible to comprehensively evaluate the distribution of haplogroup M31 and to clearly distinguish whether the aboriginal Andamanese were originated from India or Southeast Asia (Endicott et al., 2006; Reddy et al., 2007; Barik et al., 2008).

To comprehensively understand the distribution pattern of haplogroup M31 and further shed more light on the origin of the aboriginal Andamanese, we have screened 14 402 individuals (5180 from this study and 9222 from the literature), sampled from Myanmar, India, Nepal, Bhutan, Bangladesh, China, Vietnam, Thailand, Cambodia, Indonesia and Malaysia, respectively (Supplementary Table 1 and unpublished data), with especial attempt to pinpoint all potential M31 mtDNAs in the obtained mtDNA datasets so as to comprehensively evaluate the phylogeographic distribution of this haplogroup. As a result, a total of 113 mtDNAs belonging to haplogroup M31 were identified and analyzed in this study, which comprise 13 samples distilled from this study and 100 mtDNAs pinpointed from the literature. To further understand the phylogeny of haplogroup M31, additional seven mtDNAs were chosen for entire genome sequencing here.

2. Material and methods

2.1. Sampling

Totally, blood samples of 846 unrelated individuals from 14 ethnic populations across Myanmar were collected with informed consent. Total DNA was extracted by standard phenol/chloroform method. To pinpoint more M31 lineages, additional 5180 unrelated individuals from South Asia subcontinent were collected and screened for the haplogroups M31 and M32 in this study as well (Supplementary Table 1 and unpublished data).

2.2. DNA amplification and sequencing

The entire mtDNA control region for all the individuals under study were amplified, sequenced and edited as described in our previous work (Yao et al., 2003). Among the individuals belonging to haplogroup M31 pinpointed in this study, seven representatives were selected for entire mtDNA genome

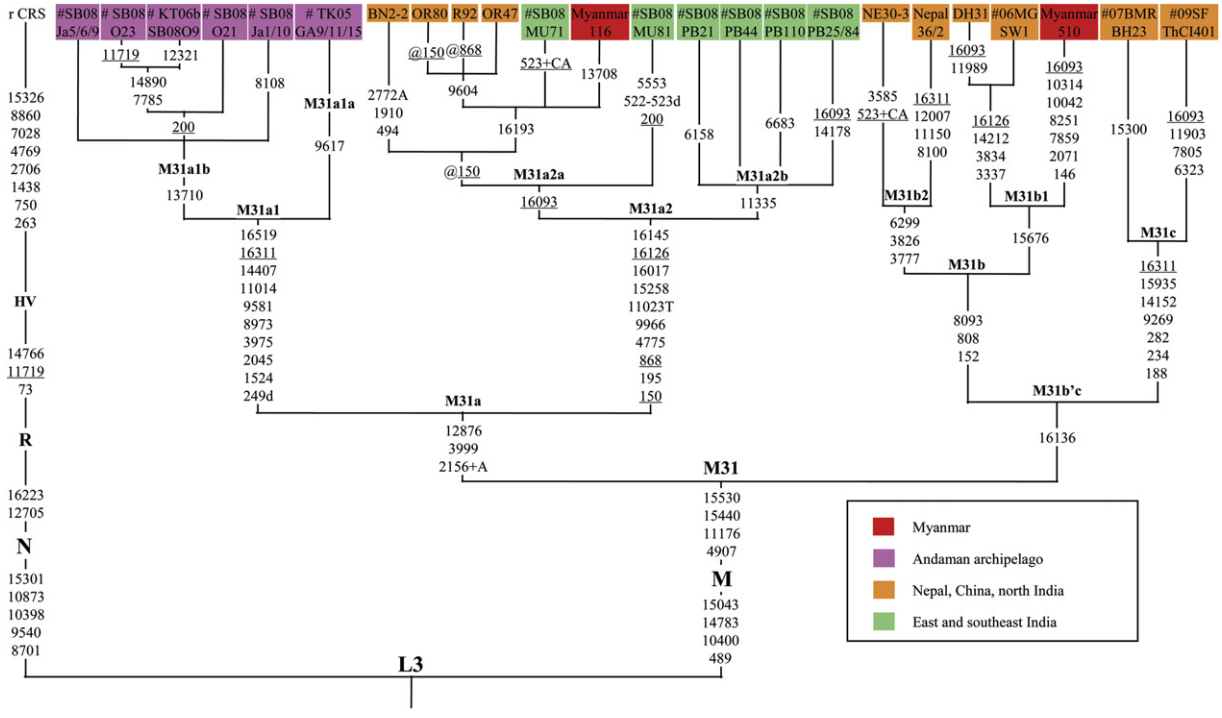

Fig. 1. Phylogenetic tree of haplogroup M31. This tree was reconstructed by adding the seven new entire mtDNA genomes obtained in this study. The designations of entire mtDNA genomes from Myanmar were marked in red, whereas the individuals from South Asian continent were labeled in pink, orange and green according to the sampling location. Mutations are recorded relative to the revised Cambridge reference sequence (rCRS; Andrews et al., 1999). Suffixes A and T indicate transversions, a plus (+) signs an insertion; recurrent mutations are underlined, and "@" highlights back mutation. Length polymorphisms (e.g., 309 + C, 309 + CC, 315 + C and 315 + CC) are ignored. The reconstruction of highly recurrent mutations (e.g., 16519 and the insertion/deletion of "CA" repeat in region 514−523) is tentative at best. Prefixes "#TK05," "#MGS06," "#KT06," "#BMR07," "#SB08," "#SF09" refer to reference information of the mtDNA genomes: Thangaraj et al. (2005), Palanichamy et al. (2006), Thangaraj et al. (2006), Reddy et al. (2007), Barik et al. (2008) and Fornarino et al. (2009), respectively.