

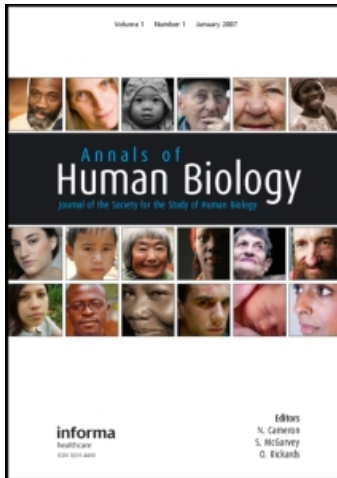
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YAP insertion signature in South Asia

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SHORT REPORT

YAP insertion signature in South Asia

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Abstract

A total of 2169 samples from 21 tribal populations from different regions of India were scanned for the Y-chromosome Alu polymorphism. This study reports, for the first time, high frequencies (8–65%) of Y Alu polymorphic (YAP) insertion in northeast Indian tribes. All seven Jarawa samples from the Andaman and Nicobar islands had the YAP insertion, in conformity with an earlier study of Andaman Islanders. One isolated case with haplotype E* was found in Dungri Bhill, a western Indian population, while YAP insertion in northeast India and Andaman tribes was found in association with haplotype D* (M168, M174). YAP insertion frequencies reported in the mainland Indian populations are negligible, according to previous studies. Genetic drift may be the causative factor for the variable frequency of the YAP insertion in the mainland populations, while the founder effect may have resulted in the highest incidence of haplotype D among the Andaman Islanders. The results of YAP insertion and the evidence of previous mtDNA studies indicate an early out of Africa migration to the Andaman and Nicobar Islands. The findings of YAP insertion in northeast Indian tribes are very significant for understanding the evolutionary history of the region.

Keywords: *Y-chromosome, alu polymorphism, Andaman islands, India*

Introduction

The human Y Alu polymorphic (YAP) element is of particular interest in the study of human evolution due to its location in the non-recombinant region of the human Y chromosome (Hammer 1994). The frequencies of Y chromosomes carrying the YAP insertion element vary greatly among human populations from different geographic locations (Hammer 1994; Hammer and Horai 1995; Hammer et al. 1997; Karafet et al. 1997). YAP insertion chromosomes appear at low frequency (<10%) in some Asian, Oceania and Amerindian

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populations, at an intermediate frequency (11–25%) in Western Eurasians, but at relatively high frequency (>30%) in Japanese, Tibetans and several African populations (Hammer 1994; Spurdle et al. 1994; Hammer and Horai 1995; Altheide and Hammer 1997; Bianchi et al. 1997; Karafet et al. 1997; Hammer et al. 1998; Thomas et al. 1998; Cruciani et al. 2002). The overall high frequency of YAP insertion chromosomes in sub-Saharan African populations led to the proposition that the original YAP insertion event occurred on an African Y chromosome and subsequently spread to other continents (Hammer et al. 1994; Underhill et al. 2001). In contrast, Hammer et al. (1997, 1998) and Altheide and Hammer (1997) hypothesized an alternative Asian origin. The lineage that acquired the YAP insertion polymorphism is divided into two sub-clusters – haplogroup E, found today in Africa and the Mediterranean, and haplogroup D, found in Japan and south-east Asia (Underhill et al. 2001). Y-chromosome data on Indian populations reveal that the parental lineages of present-day populations are predominantly derived from the original Indian gene pool (Cordaux et al. 2004; Sengupta et al. 2006) rather than a recent influx. To elucidate the regional evolutionary history, we examined the YAP polymorphism in 21 Indian tribal populations.

Materials and methods

A total of 2169 blood samples of 21 tribal populations from different regions of India (Table I) were collected and scanned for the YAP insertion following Hammer and Horai (1995). Further, the YAP insertion samples were typed with M174 and M96 primers (Underhill et al. 2000) to ascertain the haplogroup D or E affiliation.

Table I. Y chromosome haplogroup frequency.

| Region | Population | Ethnic/Linguistic category | Sample size | YAP insertion | Haplogroup D | Haplogroup E |
|-----------------------------|------------------|----------------------------|-------------|---------------|--------------|--------------|
| Northeast India | Dirang Monpa | Tibeto-Burman | 50 | 17 | 17 (34%) | 0 |
| | Shertukpen | Tibeto-Burman | 50 | 4 | 4 (8%) | 0 |
| | Lepcha | Tibeto-Burman | 50 | 8 | 8 (16%) | 0 |
| | Sonawal Kacharia | Tibeto-Burman | 112 | 0 | 0 | 0 |
| | Wanchu | Tibeto-Burman | 125 | 0 | 0 | 0 |
| | Lachungpa | Tibeto-Burman | 51 | 33 | 33 (65%) | 0 |
| Eastern India | Pauri Bhuiya | Austro-Asiatic | 120 | 0 | 0 | 0 |
| | Munda | Austro-Asiatic | 102 | 0 | 0 | 0 |
| | Toto | Tibeto-Burman | 102 | 0 | 0 | 0 |
| Central India | Kamar | Dravidian | 111 | 0 | 0 | 0 |
| | Malpaharia | Austro-Asiatic | 114 | 0 | 0 | 0 |
| | Hill Kolam | Dravidian | 123 | 0 | 0 | 0 |
| | Madia | Dravidian | 140 | 0 | 0 | 0 |
| | Kathodi | Indo-European | 120 | 0 | 0 | 0 |
| | Andh | Dravidian | 115 | 0 | 0 | 0 |
| Southern India | Jenukuruba | Dravidian | 114 | 0 | 0 | 0 |
| | Betta Kuruba | Dravidian | 115 | 0 | 0 | 0 |
| Western India | Dungri Bhill | Indo-European | 70 | 1 | 0 | 1 (1.4%) |
| Andaman and Nicobar islands | Carnicobarese | Austro-Asiatic | 114 | 0 | 0 | 0 |
| | Jarawa | Negrito/Andamanese | 7 | 7 | 7 (100%) | 0 |
| Total | | | 2169 | 73 | 72 | 1 |

Results and discussion

Out of 2169 samples, the YAP insertion was observed in 73 samples (Table I) of which only one (1.4%) YAP insertion sample from Dungri Bhill of Gujarat belonged to haplogroup E. This can be ascribed to a recent admixture from an African or Middle Eastern population. The Shia Muslims of north India, who trace their origin to the Middle East, exhibit the presence of YAP element with haplogroup E lineage at a frequency of 11% (Agrawal et al. 2005). Seventy-two YAP insertion chromosomes possessed the M174 mutation classified under haplogroup D comprising the Dirang Monpa (34%), Shertukpen (8%), Lepcha (16%), Lachungpa (65%) and Jarawa (100%) tribes. Our results on Jarawa are in conformity with the findings of Thangaraj et al. (2003). It is well established that the Andaman Islanders (Onge, Great Andamanese and Jarawa) are the extant Negrito groups in south Asia who share a common cultural background including language, in addition to mtDNA lineages and Y-chromosome haplotype D*, and have closer ties with Asians than Africans (Thangaraj et al. 2003, 2005; Palanichamy et al. 2006). Haplotype D* is also found in central Asia (Karafet et al. 2001). The phylogenetic order of YAP lineages D* and E* is still uncertain. Presence of paragroup DE* among five Nigerians led Weale et al. (2003) to dissect the interior branching order of the YAP lineages and opine that it is impossible to impute the origin of the YAP clade with certainty. Regardless of the branching order of DE*, D*, and E*, the view that male Andaman Islanders descended from Asian colonizers needs further scrutiny.

Evolutionary implications

All Y chromosomes that are not exclusively African have M168 mutation. The M168 lineage evolved into three distinct sub-clusters: One with the Alu insertion, YAP (DE haplogroup) and the other two lineages, C (RPS4Y/M216) and F* (M89/M213). Underhill et al. (2001) suggested that an African population with M168 mutation dispersed from the Horn of Africa via a coastal or interior route about 50 000–45 000 years ago (Walter et al. 2000) towards southern Asia, where the C lineage (RPS4Y/M216 mutations) probably originated. The YAP insertion probably occurred on an Asian Y chromosome as long ago as ~55 000 years (Hammer et al. 1998) based on the evidence of ancestral alleles for M40 and M96 on exclusively Asian M174 chromosomes (Altheide and Hammer 1997). The ancestral allele of M174 found exclusively in Africa, supports an African origin of YAP insertion (Underhill 2001) but the time of mutational events on the Asian YAP insertion chromosome (Hammer et al. 1998) gives antiquity to M174. Our findings of the presence of the YAP insertion in northeast Indian tribes and Andaman islanders with haplogroup D indicate that some of the M168 chromosomes have given rise to the YAP insertion and M174 mutation in south Asia. The presence of C*, YAP insertion and F* in India (Kivisild et al. 2003; Cordaux et al. 2004; Sengupta et al. 2006; Thangaraj et al. 2003) suggests that the Y chromosome is well differentiated into major lineages in south Asia. Then they moved towards southeast Asia and the Andaman Islands. Andamanese maternal links have been established through mtDNA M31 lineage with the eastern part of India in the Rajbansi of West Bengal (Palanichamy et al. 2006) and the Pauri Bhuiya of Orissa (our unpublished data). After reaching the southern part of East Asia descendants of the initial dispersal, led to a northward diaspora thus peopling across all of East Asia (Su et al. 1999). Some of the YAP insertion chromosomes without the M174 mutation reached the Mediterranean via Central Asia and gave rise to the E lineage with mutations at M40 and M96 (~31 000 years ago; Hammer et al. 1998). This E lineage back-migrated to Africa through the Levant as

hypothesized by Hammer et al. (1997) and Altheide and Hammer (1997). It is also evident that haplotype E-M34 chromosomes were probably introduced into Ethiopia from the Near East (Cruciani et al. 2004). The hypothesis of a back migration from Asia to Africa is strongly supported by the current phylogeography of the Y-chromosome variation, because haplogroup K2 and paragroup R1b*, both belonging to the otherwise Asiatic macro haplogroup K, have only been observed at high frequencies in Africa (Cruciani et al. 2002; Luis et al. 2004). Thus the major sub-sets of Y lineages that arose from the M168 lineage do not trace to an African origin. Likewise the M, N and R haplogroups of mtDNA have no indication of an African origin. In the light of recent findings by Olivieri et al. (2006) the scenario of a back migration into Africa is supported by two features of mtDNA: M1 (with an estimated coalescence time of 38.6 ± 7.1 ky) and U6 (with an estimated coalescence time of 45.1 ± 6.9 ky), which are predominantly north African clades arose in southwestern Asia and differentiated into their major sub-clades while they were in the Mediterranean area and only later some sub-sets of M1a (with an estimated coalescence time of 28.8 ± 4.9 ky), U6a2 (with an estimated coalescence time of 24.0 ± 7.3 ky) and U6d (with an estimated coalescence time of 20.6 ± 7.3 ky) diffused to East and North Africa through the Levant. Thus modern humans used a southern coastal route for their 'Out of Africa' exit, and the Levantine route from Asia to Africa for 'back migration'.

In conclusion, haplogroup D exhibits a very distinct spatial pattern. The 100% incidence of YAP insertion (haplogroup D*) in the Jarawas is due to a founder effect inferred from known demographic history (Thangaraj et al. 2003). The presence of high frequency of the YAP insertion in specific northeast Indian tribes may be due to genetic drift. Otherwise the YAP insertion is not observed in the Indian populations except for a meagre 0.6% among the Koraga tribe of Karnataka in south India (Cordaux et al. 2004). The M168 chromosomes may have taken a southern route for an initial out of Africa migration to south Asia, later being well differentiated into the major sub-sets in south Asia. One such sub-set with YAP insertion haplotype reveals that the Andaman Islanders may be one of the oldest population groups. The findings of similar footprints in northeast Indian tribes is very significant for understanding regional evolutionary history in the light of the recent findings of Olivieri et al. (2006).

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