Gene Flow from the Indian Subcontinent to Australia: Evidence from the Y Chromosome

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Summary

Phenotypic similarities between Australian Aboriginal People and some tribes of India were noted by T.H. Huxley during the voyage of the Rattlesnake (1846–1850) [1]. Anthropometric studies by Birdsell [2] led to his suggestion that a migratory wave into Australia included populations with affinities to tribal Indians. Genetic evidence for an Indian contribution to the Australian gene pool is contradictory; most studies of autosomal markers have not supported this hypothesis [3–5]; [6] and references therein). On the other hand, affinities between Australian Aboriginal People and southern Indians were suggested based on maternally inherited mitochondrial DNA [6]. Here, we show additional DNA evidence in support of Huxley’s hypothesis of an Indian-Australian connection using single-nucleotide polymorphisms (SNPs) and short tandem repeats (STRs) on the nonrecombining portion of the Y chromosome (NRY). Phylogenetic analyses of STR variation associated with a major Australian SNP lineage indicated tight clustering with southern Indian/Sri Lankan Y chromosomes. Estimates of the divergence time for these Indian and Australian chromosomes overlap with important changes in the archaeological and linguistic records in Australia. These results provide strong evidence for an influx of Y chromosomes from the Indian subcontinent to Australia that may have occurred during the Holocene.

Results and Discussion

Approximately 50% [7, 8] of the Y chromosomes in aboriginal Australians have a C → T transition [9] at position 711 in the RPS4Y gene (RPS4Y711, C → T). This transition at RPS4Y (as well as a phylogenetically equivalent mutation: M216) defines haplogroup C (Hg C), one of five major haplogroups that form the base of a highly resolved evolutionary tree of NRY lineages [10–13]. Figure 1 shows an abbreviated form of this tree. Hg C chromosomes are found at varying frequencies in Asian, Native American, and Pacific populations; however, they are absent or at low frequency in Europe and Africa [7, 10, 12]. In Australia, Hg C chromosomes have been found to be associated with a 16-bp deletion at the DYS390 STR locus (DYS390.1) [14], creating short alleles with 18–20 repeats. Chromosomes carrying short DYS390 alleles were found in 110 of 220 individuals sampled from Arnhem Land, the Northern Territories, the Kimberleys, and the Great Sandy Desert [8, 14, 15]. To investigate the origins of aboriginal Australian Hg C chromosomes, we expanded our previous SNP surveys [7, 12] by genotyping RPS4Y711, C → T in a total of 3,656 individuals from 52 worldwide populations (Figure 2), including the Warlpiri tribe from central Australia [16], and 592 Y chromosomes from several localities in southern India [17] and Sri Lanka (hereafter defined as the Indian subcontinent). We found 433 Hg C chromosomes in 30 populations (Figure 2). To further differentiate Hg C chromosomes, we genotyped four SNPs associated with the RPS4Y711, T allele: M216, M217, M38, and M8 [10, 11]. Figure 1 depicts the evolutionary relationships of Hg C and its subtypes: haplogroups C1, C2, and C3. Chromosomes that have the ancestral state at the M217, M38, and M8 mutational sites, and the derived state at both RPS4Y711, and M216, are designated “C*”. The asterisk represents an interior node on the tree [13]. Approximately 20% of the Hg C chromosomes in our survey were C*. While C* chromosomes were present in combination with C3 chromosomes in mainland southeast Asia, and with C2 chromosomes in east Indonesia, coastal Papua New Guinea (PNG), and Micronesia, C* chromosomes were the only Hg C chromosomes present in our Indian subcontinent and aboriginal Australian samples (Figure 2). All other populations lacked C* chromosomes.

Does this sharing of C* chromosomes support Huxley’s hypothesis [1] of an Indian-Australian connection? One complicating factor is the possibility that C* chromosomes are actually a heterogeneous mixture of derived haplogroups that are distinct in Indian subcontinent and Australian populations (i.e., marked by derived states at different SNPs yet to be discovered). To directly examine the evolutionary origins of C* chromosomes, we genotyped 10 Y-linked STRs in a sample of 63 C* chromosomes and 64 C1, C2, and C3 chromosomes. Figure 3 shows a median-joining (MJ) network [18] constructed from SNP and STR variation associated with these chromosomes. The MJ network rooted among C* chromosomes from the Indian subcontinent. The Indian subcontinent C* chromosomes were widely distributed across the central portion of the network, while the aboriginal Australian chromosomes formed a tight cluster
C chromosomes (6.88) is less than the number of mutational steps within those from the Indian subcontinent (7.32). Finally, the lowest average number of pairwise differences among haplotypes (6.88) in Table 1 is between aboriginal Australian and Indian subcontinent haplotypes rather than between aboriginal Australian chromosomes and those of their close geographic neighbors in Melanesia.

If we are, indeed, detecting a genetic connection in accord with Huxley’s hypothesis, it would be interesting to estimate the timing of divergence between Australian and Indian C* chromosomes. To explore this further, we used the STR data to estimate divergence times using two published methods. In both methods, we assumed a generation time of 25 years. Furthermore, we assumed a mutation rate of $2.08 \times 10^{-3}$ that is based on the average rate from three pedigree studies [19–21] and included 8,169 meioses in 9 Y-STR loci. In the first dating method, the $(\bar{\mu}^2)^2$ genetic distance was selected because it provides a robust divergence estimate that remains linearly with increasing time and it is largely independent of population size [22]. To calculate 95% CIs, 1,000 bootstrap samples were performed to obtain the standard errors of $(\bar{\mu}^2)$. The average $(\bar{\mu}^2)$ divergence between Indian and Australian C* chromosomes was $0.256 \pm 0.105$. Thus, the age of the divergence is 62 generations (95% CI = 12–111 generations) or 1,550 years (95% CI = 300–2,775 years). The variance displayed in these 95% CIs does not include the uncertainty in the mutation rate. Second, we used a Bayesian method for estimating the time to the most recent common ancestor (TMRCA) described in Walsh [23]. We used this method to calculate 95% CIs for the TMRCA using tallies of the number of STR differences between populations of Hg C chromosomes (Table 1) are consistent with the aforementioned phylogenetic inferences. First, Indian subcontinent chromosomes are more diverse than aboriginal Australian chromosomes. Second, the average number of mutational steps between Indian subcontinent and aboriginal Australian Hg C chromosomes (6.88) is less than the number of mutational steps within those from the Indian subcontinent (7.32). Finally, the lowest average number of pairwise differences among haplotypes (6.88) in Table 1 is between aboriginal Australian and Indian subcontinent haplotypes rather than between aboriginal Australian chromosomes and those of their close geographic neighbors in Melanesia.

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Figure 3. Median-Joining Network of Hg C Chromosomes
The tree was constructed from STR and SNP variation with NETWORK3.1 (http://www.fluxus-engineering.com). The MJ network was rooted with 12 Hg B chromosomes from sub-Saharan Africa. Each circle represents a unique STR haplotype, and the size of the circle is proportional to the number of individuals sharing that haplotype. Haplotypes were constructed from the following STRs: DYS19, DYS391, DYS393, DYS385A, DYS385B, DYS426, DYS388, DYS392, DYS389I, DYS389II, and DYS439. DYS390 was omitted because it violates the single-step mutation model. For all analyses, we subtracted DYS389I from DYS389II because the latter contains the former. SNPs were given a weight of 20, and STRs were given a weight of 1. Dotted lines encompass Hg C subtypes (C1–C3) as well as the internal node C*. The double hatched line indicates that the length of the branch is not to scale. Color codes for each population group are shown in the key (the white circle represents a single Uygur from Central Asia). Populations are defined in Figure 2, except OCE, which includes Melanesians, Polynesians, and Micronesians, and ISC, which includes southern Indians and Sri Lankans.

Table 1. Haplogroup C STR Diversity within and between Population Regions

<table>
<thead>
<tr>
<th>Sample</th>
<th>AAP</th>
<th>ISC</th>
<th>SEA</th>
<th>EIN</th>
<th>MEL</th>
<th>POL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Aboriginal Australian People (AAP)</td>
<td>4.10</td>
<td>6.88</td>
<td>9.33</td>
<td>9.47</td>
<td>8.94</td>
<td>8.99</td>
</tr>
<tr>
<td>Indian subcontinent (ISC)</td>
<td>0.256</td>
<td>7.32</td>
<td>8.89</td>
<td>11.45</td>
<td>10.76</td>
<td>10.11</td>
</tr>
<tr>
<td>Southeast Asians (SEA)</td>
<td>0.459</td>
<td>0.107</td>
<td>7.12</td>
<td>11.98</td>
<td>12.34</td>
<td>13.75</td>
</tr>
<tr>
<td>East Indonesians (EIN)</td>
<td>0.653</td>
<td>1.265</td>
<td>1.294</td>
<td>9.82</td>
<td>11.71</td>
<td>13.10</td>
</tr>
<tr>
<td>Melanesians (MEL)</td>
<td>0.501</td>
<td>0.808</td>
<td>1.056</td>
<td>0.808</td>
<td>9.47</td>
<td>9.31</td>
</tr>
<tr>
<td>Polynesians (POL)</td>
<td>0.886</td>
<td>0.769</td>
<td>1.277</td>
<td>1.778</td>
<td>0.393</td>
<td>3.34</td>
</tr>
</tbody>
</table>

The average number of pairwise differences are shown within (P_{iX} along diagonal) and between populations (P_{iXY} above diagonal); pairwise ($\bar{\theta}_{iX}^2$) genetic distance values are shown below the diagonal. Values were computed using ARLEQUIN [33] and MICROSAT [34]. AAP – 14 Central Desert, 5 Great Sandy Desert, and 2 Western Australians; ISC – 16 Southern Indians and 3 Sri Lankans; SEA – 12 Yao, 11 Tuja, 3 Miao, 3 Vietnamese, and 1 Malay; EIN – 10 Moluccans, 8 Nusa Tenggarans, and 1 Javan; MEL – 10 Vanuatuans and 5 Papua New Guineans; and POL – 13 Samoans, 8 Tahitians, and 3 Tongans.
ctesty of some aboriginal Australian and Indian subcontinental C* chromosomes. Moreover, the fact that aboriginal Australian chromosomes represent a subset of the C* diversity found in the Indian subcontinent may provide evidence that the Australian C* chromosomes descended from an Indian subcontinental ancestor(s). Indeed, the positions of Indian subcontinental C* lineages in Figure 3 (i.e., basal to many of the derived haplotypes) lends support to the hypothesis of an Indian origin of the RPS4Y deleterion, albeit with the spread of C* chromosomes to or within Australia.

The absence of the DYS390.1 deletion in the Indian subcontinental sample may imply that the deletion event occurred either in India, on a Y chromosome that was migrating from India to Australia, or in Australia well after the migration of C* chromosomes from India. Further studies are needed to distinguish among these hypotheses and to infer the evolutionary forces that resulted in such high frequencies of C* chromosomes carrying the DYS390.1 deletion in contemporary populations of Australian Aboriginal People.

In sum, we found that 50% of the Y chromosomes sampled from aboriginal Australians share common ancestry with a set of Y chromosomes that represent less than 2% of the sampled Indian subcontinental paternal gene pool. The similarity among C* chromosomes is unlikely to have been caused by chance convergence because we genotyped ten independent STRs. The observed pattern is not specific to central Australians, since our sample also included individuals from the Great Sandy Desert and from Western Australia, and our estimate of the frequency of C* chromosomes agrees remarkably well with other studies of greater numbers of aboriginal Australian Y chromosomes in Arnhem Land, the Great Sandy Desert, the Kimberleys, and the Northern Territory [8, 14, 15].

A southern route of migration from Africa to Australia in the Pleistocene has often been suggested to explain similarities among some populations in Africa, India, Southeast Asia, and Australia [4, 5]. The presence of closely related C* chromosomes in populations sampled from the tropical corridor between south Asia and the Pacific is consistent with this hypothesis (Figure 2). However, our analyses suggest a mid-Holocene common ancestry of aboriginal Australian and Indian subcontinental C* chromosomes. The divergence times reported here correspond with a series of changes in the Australian anthropological record between 5,000 years ago and 3,000 years ago, including the introduction of the dingo [24]; the spread of the Australian Small Tool tradition [25]; the appearance of plant-processing technologies, especially complex detoxification of cycads [26]; and the expansion of the Pama-Nyungan language over seven-eighths of Australia [27]. Although there is no consensus among anthropologists, the former three changes may have links to India, perhaps the most relevant of which is the introduction of the dingo, whose ocean transit was almost certainly on board a boat. In addition, Dixon [28] noted some similarities between Dravidian languages of southern India and Pama-Nyungan languages of Australia.

The combined genetic (Y chromosome and mitochondrial DNA) and anthropological evidence supports Holocene contact between the Indian subcontinent and Australia, although there is a need for more research in this area. Genetic dating of the divergence between Australian and Indian C* chromosomes will improve as the number of loci typed increases [23], as locus/allele-specific mutation rates are refined [29], and as simulation methods are developed that consider the effects of including only subsets of chromosomes sampled in different populations [30]. It is important to emphasize that Australian and Indian non-Hg C chromosomes did not show close affinities and that paternal variation cannot be used to infer the complete genetic history of Australian populations. On the other hand, weak signals for population relationships based on autosomal data may be magnified on the NRY as a result of its lack of recombination and susceptibility to higher levels of genetic drift. Therefore, the NRY can be an important tool for reconstructing the history of human populations.

Acknowledgments

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