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RESEARCH ARTICLE

Maori Origins, Y-Chromosome Haplotypes and Implications for Human History in the Pacific

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An assessment of 28 pertinent binary genetic markers on the non-recombining portion of the Y chromosome (NRY) in New Zealand Maori and other relevant populations has revealed a diverse genetic paternal heritage of extant Maori. A maximum parsimony phylogeny was constructed in which nine of the 25 possible binary haplotypes were observed. Although ~40% of the samples have haplotypes of unequivocal European origin, an equivalent number of samples have a single binary haplotype that is also observed in Indonesia and New Guinea, indicative of common indigenous Melanesian ancestry. The balance of the lineages has either typical East Asian signatures or alternative compositions consistent with their affinity to Melanesia or New Guinea. Molecular analysis of mtDNA variation confirms the presence of a single predominant characteristic Southeast Asian (9-bp deletion in the Region V) lineage. The Y-chromosome results support a pattern of complex interrelationships between Southeast Asia, Melanesia, and Polynesia, in contrast to mtDNA and linguistic data, which uphold a rapid and homogeneous Austronesian expansion. The Y-chromosome data highlight a distinctive gender-modulated pattern of differential gene flow in the history of Polynesia. Hum Mutat 17:271–280, 2001.

KEY WORDS: SNP; DHPLC; Maori; Y-chromosome haplotypes; mitochondrial DNA; NRY; Polynesian origins; admixture; sex-specific gene flow

DATABASES:
http://www.ncbi.nlm.nih.gov/SNP/ (dbSNP/NCBI); http://hpgl.stanford.edu/ (Stanford University Human Population Genetics Laboratory)

INTRODUCTION

Considerable multidisciplinary evidence suggests that the settlement of Aotearoa (New Zealand) occurred about 800–1,000 years ago by Polynesian ancestors who trace their origins to the central Eastern Pacific [Sutton, 1994], perhaps more specifically to the Cook Islands [Bellwood, 1989]. These migrations are a part of the overall colonization of Austronesian Polynesia that started from Southeast Asia about 3,000–4,000 years ago. While much of the evidence pertaining to the origins of Polynesians is archeological and linguistic [Bellwood, 1978; Gray and Jordan, 2000], mitochondrial DNA evidence has provided an independent assessment that is consistent with a Southeast Asian Austronesian influence [Hertzberg et al., 1989; Murray-McIntosh et al., 1998;

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Merriwether et al., 1999]. A different scenario, recently suggested by Y-chromosome markers, indicates that Melanesian populations strongly influenced the Polynesian gene pool [Kayser et al., 2000; Capelli et al., 2001]. Europeans began to settle in New Zealand at the beginning of the 19th century, and by 1850 they already outnumbered Maori, who today represent only about 13% of the total New Zealand population. The settlement of Europeans has not only influenced the environment of Aotearoa and Maori culture, but also the Maori gene pool. This has complicated the interpretation of molecular genetic evidence concerning the topic of Maori origins. The sequential accumulation of binary markers associated with the non-recombining portion of the Y chromosome (NRY) permits construction of stable compound haplotypes [Jobling and Tyler-Smith, 1995]. These haplotypes unequivocally identify lineages with shared common ancestry, which often display population specificity due to a reduction of variation relative to other loci, including mtDNA [Shen et al., 2000]. These properties make Y-chromosome haplotypes highly informative for the inference of population origins and migration patterns. While NRY associated PCR compatible polymorphisms are providing independent genetic evidence regarding the origins of Polynesians [Hurles et al., 1998; Su et al., 2000a; Kayser et al., 2000], limited data exist for Maori. Two Y-chromosome restriction length polymorphisms (RFLPs), a rearrangement polymorphism and an Alu polymorphism have been studied in 33 Maori [Spurdle et al., 1994]. These initial lower resolution results indicated that the Maori have a closer affinity with "Caucasoids" than they do with Africans. Here we attempt to illuminate the origins of the Maori by using a combination of high resolution Y-chromosome binary haplotypes and mtDNA sequence variation. In addition, we investigate whether evidence exists for sex specific modulated gene flow, as has been shown in other populations [Bamshad et al., 1998; Passarino et al., 1998; Zerjal et al., 1997].

MATERIAL AND METHODS

DNA Samples

The geographic locations of the various populations studied are shown in Figure 1. The respective Institutional Review Boards and Human Subjects Committees approved all sampling pro-

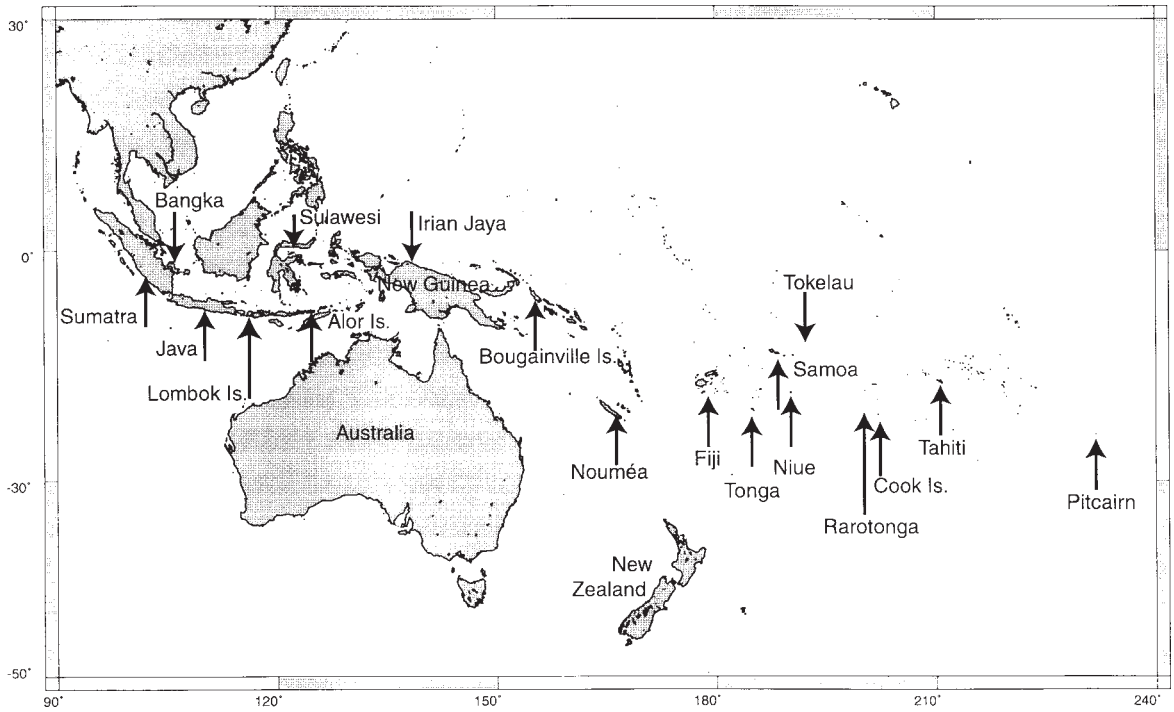

FIGURE 1. **Geographic locations of the population samples.**

tocols used in the survey. A total of 148 human DNA samples from Victoria University were included in the mtDNA survey. Informed consent was obtained from all volunteers under the supervision of the Wellington Ethics Committee. The NRY survey included 71 males, 54 of which were of stated Maori heritage. The remaining 17 males included seven from Samoa, three Tonga, one Rarotonga, one Moumea, one Tokelau, one Cook Islands, and three from unspecified locations in Polynesia. The 77 females are described as follows: 59 Maori, four Fiji, five Samoa, one Tokelau, one Cook Island, one Pitcairn, two Niue, and four unspecified Polynesia. A total of 17 male samples from Indonesia (all having the derived allele for RPS4YC711T) were included in the NRY analysis. These included four from Alor, two Java, three Irian Jaya, four Sulawesi, one Sumatra, one Bangka Island, and two Lombok Island. Additional NRY data [Underhill et al., 2000] involving samples from Australia (n = 7), New Guinea (n = 15) and Bougainville Island (n = 8) are discussed.

Y-Chromosome Binary Polymorphism PCR and Haplotyping

The twenty-eight binary markers included four taken from the literature (see below) and 24 detected by denaturing high-performance liquid chromatography (DHPLC) [Oefner and Underhill, 1998]. The DHPLC technique (see below) was used to genotype the 24 DHPLC identified markers as well as RPS4YC711T [Bergen et al., 1999] and SRY9138 [Whitfield et al., 1995]. Both YAP [Hammer and Horai, 1995] and 12f2 [Casanova et al., 1985; Sun et al., 2000] were typed following recommended PCR protocols. Marker details are as described [Underhill et al., 2000, 2001] and are also available upon request. All DHPLC scored markers had a uniform annealing temperature, which allowed a single PCR protocol to be used. The thermal cycling regime used consisted of an initial denaturation at 95°C for 10 min to activate the AmpliTaq Gold® DNA polymerase, 14 cycles of denaturation at 94°C for 20 sec, primer annealing at 63 to 56°C for 30 sec using 0.5°C decrements, and extension at 72°C for 1 min, followed by 20 cycles at 94°C for 20 sec, 56°C for 1 min, 72°C for 1 min, and a final 5-min extension at 72°C. Each 50-μl PCR reaction contained 1 U of AmpliTaq Gold® DNA polymerase, 10 mM Tris-HCl, pH 8.3, 50 mM KCl, 2.5 mM MgCl2, 0.1 mM each of the four deoxyribonucleoside triphosphates, 0.2 μM each of forward/reverse primers, and 50 ng of genomic DNA. All PCR yields were determined semi-quantitatively on ethidium bromide stained agarose gels. Since limited amounts of male DNA was available, these samples were genotyped by using a top-down hierarchical approach in which polymorphisms were typed only in samples carrying mutations at higher levels.

DHPLC Analysis

Unpurified PCR products were mixed at an equimolar ratio with a similar amount of reference Y-chromosome amplicon and subjected to a 3-min 95°C denaturation step followed by gradual reannealing from 95 to 65°C over 30 min. Ten microliters of each mixture were loaded onto a DNASep™ column (Transgenomic, San Jose, CA), and the amplicons were eluted in 0.1M triethylammonium acetate, pH 7, with a linear acetonitrile gradient at a flow rate of 0.9 ml/min. Appropriate temperature conditions were predicted by computer simulation (available at http://insertion.stanford.edu/melt.html). Mismatches were recognized by the appearance of two or more peaks in the elution profiles.

Y-Chromosome Microsatellites

The microsatellites studied included DYS19 [Roewer et al., 1992], also referred to as DYS394 [Jobling and Tyler-Smith, 1995], YCAIIa, YCAIIb [Mathias et al., 1994] and DYS390 [Forster et al., 1998]. For DYS19, YCAIIa, and YCAIIb, one fluorescent labeled primer and one unlabeled primer for each locus was amplified in a 10 μl PCR reaction using reagent and PCR conditions as described in Ruiz Linares et al. [1996]. Following PCR, 1 μl of reaction product was loaded onto a standard 6% polyacrylamide denaturing gel with electrophoresis performed in an ABI 373A sequencer. Internal size standards were added to each sample to permit quantitative sizing of each fragment using GeneScan 672 software. The DYS390 locus was amplified and then sequenced directly to determine both its length and repeat motif composition. PCR products were purified with QIAGEN (Valencia,

CA) QIAquick spin columns. Subsequently, both strands were sequenced using the amplimers as sequencing primers and ABI Dye-terminator cycle sequencing reagents (Applied Biosystems, Foster City, CA). The sequencing products were purified with Centriflex™ gel filtration cartridges (Edge Biosystems, Gaithersburg, MD) and analyzed on an Applied Biosystems 373A sequencer.

mtDNA Genotyping

The presence of the 9-bp deletion in the mtDNA region V was determined by PCR, followed by gel electrophoresis with ethidium bromide visualization, as described in Wrischnik et al. [1987]. The European and Asian mtDNA haplogroups were evaluated as described in Macaulay et al. [1999].

Maximum Parsimony

A set of 28 markers were used to deduce a Y-chromosome phylogeny in which a network of branches is drawn that minimizes the number of mutational events required relating the lineages and whose sequential succession is unequivocal [Jobling and Tyler-Smith, 1995]. The root of the resulting tree was located by using orthologous allelic information from three great ape species (chimp, gorilla, and orangutan) as outgroups.

RESULTS

Y-Chromosome Haplotypes and Frequencies

A maximum parsimony phylogeny (Fig. 2) was constructed based on the 28 relevant binary markers surveyed. Nine of the 25 possible haplotypes defined in the phylogeny were detected and their frequencies are given in Table 1. The nine observed haplotypes are distributed throughout the phylogeny and indicate quite diverse ancestry.

The RPS4YC711T and M216 substitutions define a major monophyletic clade in the Y-chromosome genealogy [Bergen et al., 1999; Karafet et al., 1999; Underhill et al., 2001] that includes ht2 through ht6. Only ht3, defined by M38, was found in the Maori and Polynesian samples studied. Its frequency in the two populations was 42.6% and 41.2%, respectively. Since M38 was originally ascertained in New Guinea where its frequency is 17.4% [Underhill et al., 2000], we also assessed its frequency in 17 Indonesian samples that were characterized previously by the RPS4YC711T and M216 polymorphisms. The frequency of ht3 in these Indonesians was 58.8%. We have not observed ht3 elsewhere in Asia [Underhill et al., 2000], indicating that it marks an important lineage that is indigenous to the Pacific and is associated with Melanesian populations. The RPS4YC711T related lineage, ht6, defined by M217, was not observed in the Indonesian, Maori, or Polynesian samples studied. Neither was ht2, defined by M210 and present only in Australia, nor ht5 defined by M8, M105 and M131 and found in Japan previously [Underhill et al., 2000]. The ancestral ht4, has been observed, so far, only in Indonesia.

Haplotype ht12 is defined by M122 and occurs

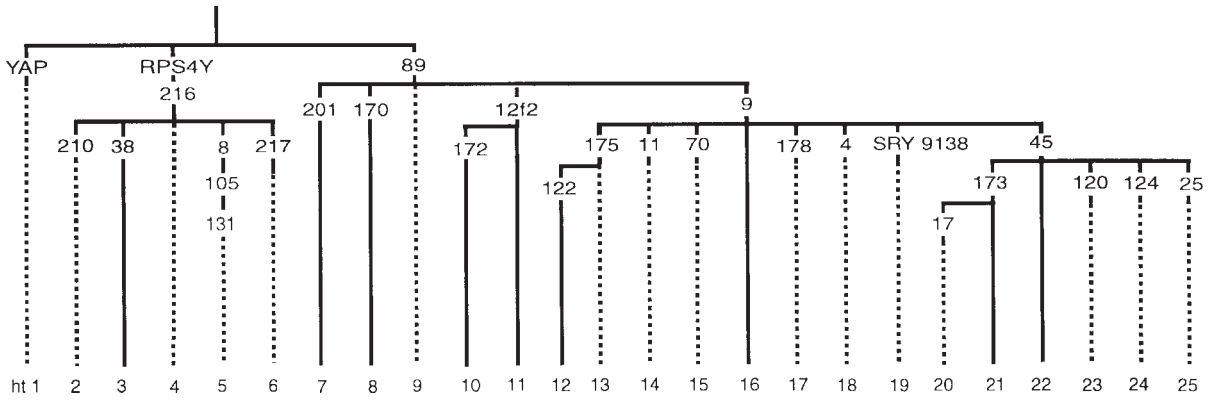

FIGURE 2. **Maximum parsimony phylogeny of 28 Y chromosome binary markers that define 25 haplotypes. The nine haplotypes observed in Maori and Polynesians are indicated by solid lines. The remaining 16 unobserved haplotypes are indicated by dashed lines.**

TABLE 1. Y Haplotype Distribution in Maori and Other Polynesians

Haplotype number	ht3	ht4	ht7	ht8	ht10	ht11	ht12	ht16	ht21	ht22
Defining marker	M38	RPS4Y	M201	M170	M172	12f2	M122	M9	M173	M45
Maori (N=54)	23	0	1	5	1	1	3	1	18	1
Other Polynesians (N=17)	7	0	0	0	1	0	3	3	3	0

An additional sample of 17 Indonesians, selected for being RPS4YT derived, was also analyzed. Ten were ht3 and 7 were ht4. In Indonesia, ht3 (RPS4Y/M216/M38) occurs in Irian Jaya, Bangka Is., S. Sulawesi, Lombak Is., S. Sumatra and E. Java. Ht4 (RPS4Y/M216) was observed in Lombok Is., Alor Is., Irian Jaya, S. and C. Sulawesi.

at 17.4% in Polynesians and 5.5% in Maori. This haplotype is very common throughout East Asia [Underhill et al., 2000, 2001], and has been associated with the expansion of agriculture and Sino-Tibetan languages from China [Su et al., 2000b].

Haplotypes ht7, 8, 10, 11, and 21 are common in Europe [Semino et al., 2000]. Taken together these five European haplotypes occur at 48.1% in Maori and 23.5% in our Polynesian samples, respectively. Haplotype ht16 is defined by the M9 mutation only and occurs at 5.5% in Maori and 5.9% in Polynesia, respectively. In New Guinea, ht16 occurs at 30.4%, while it is extremely rare elsewhere [Underhill et al., 2000]. Finally, haplotype ht22 is defined by the M45 mutation and occurs in a single Maori subject (1.9%).

Y-Chromosome Microsatellites

Table 2 presents the distribution of microsatellite haplotypes associated with each of the lineages defined by binary haplotypes (ht3, ht12, ht16) that were not considered of European heritage. The DYS390 locus, whose complex structure, composed of both tetra and trinucleotide repeats, [Forster et al., 1998] is the least variable in each of the three lineages. All the individuals belonging to the M38 lineage exhibited a short (< 200 bp) allele for the DYS390 locus, with the deletion of the DYS 390.3 element [Forster et al., 1998; Kayser et al., 2000]. The tetranucleotide repeat DYS19 and the dinucleotide repeats YCAII a and b are more variable. However, most of the haplotypes within each of the three lineages could be connected to each other by single steps, implying very low diversity within the lineages [Cooper et al., 1996].

mtDNA Variation

Table 3 lists the distribution of the mtDNA groups observed in Maori and the other Polynesians. The 9-bp deletion motif, common in Asian and Polynesian populations, occurred at 85% in Maori and in all of the Polynesian samples. The remaining 15% of mtDNAs were characteristic of European haplogroups.

DISCUSSION

We have simultaneously analyzed the paternal and maternal heritage of Maori using polymorphic Y-chromosome and mitochondrial DNA systems. Both types of uniparentally in-

TABLE 2. Microsatellite Haplotypes Linked to Binary Haplotypes ht3, ht12 and ht16 in Maori and Polynesians

	ht3			
	YCAII a/b	DYS19 bp	DYS390 bp	N
Maori (N=24)	1/1	194	199*	13
	1/1	190	199*	4
	1/1	194	195*	1
	3/1	186	199*	1
	3/1	190	199*	1
	3/1	194	199*	1
	4/1	190	199*	2
	4/1	194	199*	1
Polynesians (N=5)	1/1	194	199*	1
	3/1	194	195*	1
	3/1	194	199*	1
	3/3	194	199*	1
	4/1	194	199*	1
	ht12			
	YCAII a/b	DYS19 bp	DYS390 bp	N
Maori (N=3)	3/1	194	215	2
	4/1	194	215	1
Polynesians (N=3)	3/1	194	215	2
	4/1	194	215	1
	ht16			
	YCAII a/b	DYS19 bp	DYS390 bp	N
Maori (N=1)	3/–7	186	219	1
Polynesians (N=3)	1/–7	194	219	2
	–7/–7	194	219	1

*These alleles exhibited the deletion of the 390.3 element [Forster et al. 1998; Kayser et al., 2000]. For YCAII nomenclature see Quintana-Murci et al. [1999].

TABLE 3. **Mitochondrial DNA Types Found Among Maori and Other Polynesians**

Maori (N=116)	
9bp deletion	**85.3%**
Haplogroup H	**6.9%**
Haplogroup K	**1.7%**
Haplogroup U5	**2.6%**
Haplogroup T	**2.6%**
Haplogroup J	**0.9%**
Other Polynesians (N=32)	
9bp deletion	**100%**

herited loci revealed important, although unequal, components whose contributions can now be properly recognized. After excluding admixed European components, three Y-chromosome lineages, each defined by a Single Nucleotide Polymorphism (SNP), were present in our sample: ht3, defined by RPS4Y/M216/M38; ht16, defined by M9; and ht12 defined by M122. These three lineages have all been reported previously in other Polynesian populations [Su et al., 2000a; Kayser et al., 2000]. Given the low microsatellite diversity in each of the three lineages, they probably represent the legacy of the original founding Maori population. Interestingly, the Y binary haplotypes indicated more diversity than the mtDNA, which, after excluding the European derived haplotypes, showed that 100% of Maori exhibit the 9 bp Region V deletion [Wrishnick et al., 1987]. Previous studies of mtDNA in Maori have rarely found (about 2%) haplotypes without the deletion [Hertzberg et al., 1989; Sykes et al., 1995; Murray-McIntosh et al., 1998].

The analysis of the D loop region allowed Murray-McIntosh et al. [1998] to recognize four different haplotypes. Although these haplotypes were related to each other, given the short time elapsed since the peopling of Aotearoa (about eight centuries), the authors interpreted this result as evidence that more than one founder canoe colonized the Islands, thus supporting the oral tradition of multiple settlement. Should this interpretation be correct, our Y-chromosome data also suggest multiple arrivals. In fact, at least three different Y-chromosome lineages (two of which may have had some microsatellite diversity) may have been present among the founders, including the M9 defined haplotype, which is not very frequent in Central Polynesia (about 6–10%) [Su et al., 2000a; Kayser et al., 2000; and present data]. In addition, these lineages exhibit in Aotearoa a frequency approximating that observed in other Polynesian islands [Su et al., 2000a; present data]. This suggests that the settling of Aotearoa did not result in an exceptional reduction in inherent variability present in the original homeland population, although the microsatellites associated with M38, the most frequent lineage, show a certain reduction of variability in Maori relative to the other Polynesians we studied (Table 2). These findings corroborate earlier results from minisatellite DNA surveys on the same samples [Clark et al., 1995; Hamilton et al., 1996] and similar studies on other samples from the same populations [Flint et al., 1989].

The analysis of Y-chromosome variation among Maori not only addresses their specific origin(s), but also provides more general insights into the colonization of the Pacific that involved at least one episode during the Pleistocene including Australia, followed by a much more recent Polynesian migration [Cavalli-Sforza et al., 1994]. Different hypotheses regarding Polynesian origins have been proposed. One of the most commonly discussed includes a rapid, agriculturally driven expansion event from Taiwan via Southeast Asia referred to as the "Express train to Polynesia" [Diamond, 1988; Gray and Jordan, 2000]. Alternatively, many authors have proposed a more complex pattern of interactions between Melanesia, Southeast Asia, and Polynesia, and/or migration(s) to Polynesia starting from Melanesia [Terrell, 1988; Terrell et al., 1997; Lum and Cann, 1998; Oppenheimer, 1998]. Most linguistic data have associated Taiwan with the origin of Austronesian languages supporting the "Express train to Polynesia" model [Bellwood, 1989; Gray and Jordan, 2000]. However, Dyen [1962, 1965] proposed Melanesia as the origin of the Austronesian languages on the basis of lexico-statistical methods. Support for both models has been invoked from mtDNA data. The mtDNA 9-bp deletion motif and its derivatives have been seen as strong evidence of a rapid expansion from Southeast Asia [Sykes et al., 1995; Melton et al., 1995; Redd et al., 1995]. Alternatively, a much less frequent Melanesian attributed lineage, as well as the phylogenetic analysis of the D loop variants associated with the 9-bp deletion in Polynesia but not in Asia, have been interpreted

as evidence of Austral-Melanesian associated ancestry [Richards et al., 1998].

Our Y-chromosome haplotype results offer another perspective into Polynesian history that indicates a more diverse Melanesian component than previously revealed by mtDNA. This evidence stems primarily from lineages associated with the RSP4YC711T marker that has been previously observed in the Pacific [Karafet et al., 1999; Kayser et al., 2000]. Our analysis of the binary mutations associated with RSP4YC711T now allows us to evaluate more precisely the relationships between the widespread RSP4YC711T lineages across Asia, Oceania, and America [Bergen et al., 1999; Karafet et al., 1999]. Most of the Asian and Native American Y chromosomes carrying RSP4YC711T also have the M217 marker. The only exception was a single individual from Japan, displaying the M8/M105/M131 haplotype [Underhill et al., 2001]. The M217 mutation was not found in Australasia. In particular, while M210 seems restricted to Australia [Underhill et al., 2001], M38 (ht3) has been proposed to have arisen in the populations that colonized Papua New Guinea and Melanesia about 40,000 years ago [Underhill et al., 2001]. Support for this interpretation comes from the comparison of our results to those obtained by Kayser et al. [2000]. In fact, all M38 marked chromosomes also have the DYS390.3 deletion, which Kayser et al. [2000] interpreted as an index of ancient Melanesian heritage. An analysis of Indonesian chromosomes carrying RSP4YC711T shows that the Indonesia archipelago retains a more immediate ancestral haplotype (ht4).

The identification of ht3 defined by M38 in Maori and Polynesians (in 23/54 and 7/17 subjects respectively—Table 1) unequivocally defines an important lineage with Melanesian heritage. The occurrence of ht16 lineage, although only defined by M9, is suggestive of an additional signature from Melanesia. It has been previously observed in New Guinea at 30% [Underhill et al., 2000]. In contrast to ht3 and ht16, haplotype ht12, defined by M122, indicates a Southeast Asian ancestry (in 3/54 Maori and 3/17 Polynesians; see Table 1), possibly brought to the Pacific area after the expansion of agriculture from Asia [Su et al., 2000b].

The overall collection of Y-chromosome lineages (Table 1) indicates a pronounced influence of the "Melanesian matrix" [Terrell et al., 1997] in the Polynesian gene pool, while in contrast, the inclusion of ht12 indicates the presence of additional Southeast Asian elements. The most likely explanation is a confluence of Southeast Asian and/or Austronesian speaking people that mixed with Melanesians before colonizing Polynesia. This mixed population could be related to the Lapita culture [Cavalli-Sforza et al., 1994]. Interestingly, a comparison of mtDNA and autosomal microsatellite results [Lum et al., 1998] indicated different patterns of relationships between Asian and Pacific Island populations. Molecular data from mtDNA and Y-chromosome markers can give different pictures of population history due to the modulating effects of sex and culture. In this case two elements are conspicuous:

1. Although Y-chromosome data are often more in agreement with linguistic data [Poloni et al., 1997; Zerjal et al., 1997; Passarino et al., 1998] than the corresponding mtDNA data, the opposite pattern appears in the Pacific. Namely, the spread of Austronesian languages is more congruent with the diffusion of Southeast Asian mtDNA heritage, than with that of the most prevalent Melanesian Y-chromosome lineages observed in Polynesia.
2. The Y chromosome has shown less variability than mtDNA [Shen et al., 2000]. In the case of Pacific Island populations however, Y chromosomes have more variability than mtDNA and reveal genetic signatures from both Melanesia and Southeast Asia.

These peculiarities are probably reflective of the history of settlement. It is possible that the early population that arose from the mixing of Southeast Asians and Melanesians spoke an Austronesian language. Differences in sex ratio during successful colonization and perhaps subsequent exploration may explain the observation of greater Y chromosome than mtDNA variety [Lum et al., 1998]. Specifically there may have been more unrelated males per voyage than females. In addition, there may have been a dis-

proportionate amount of non-Polynesian male mediated gene flow, compared to females, from Melanesian related populations following initial settlement. Thus, in contrast to most of mtDNA and linguistic data [Merriwether et al., 1999; Gray and Jordan, 2000], the scenario suggested here by Y-chromosome variation is more consistent with a deep genetic contribution of Melanesian ancestry to the Polynesian gene pool than with the "Express Train to Polynesia" model.

Finally, some details concerning European gene flow into the Maori gene pool can be recognized. First, our study shows that male mediated gene flow has been more prevalent than female mediated gene flow, as previously noticed throughout Polynesia [Spurdle et al., 1994; Hurles et al., 1999]. In addition, since the composition of Y-chromosome variation in Europe has been recently categorized [Semino et al., 2000], we were able to infer that European haplotypes present among Maori are typical of northwestern Europe, thus consistent with recorded history of European colonization of the Pacific.

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