POLYNESIAN ORIGINS AND MIGRATION: THE STORY ACCORDING TO NUCLEAR AND MITOCHONDRIAL DNA MARKERS

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INTRODUCTION

Genetic (DNA) studies have contributed to our understanding of Polynesian origins, ethnic affinities and migratory movements. DNA analyses may be obtained from nuclear or mitochondrial markers. The present study describes an extensive characterisation in Polynesians of nuclear DNA markers (alpha globin gene cluster - chromosome 16p13.3; beta globin gene cluster - chromosome 11p15; phenylalanine hydroxylase gene - chromosome 12q22-q24.1) as well as a 9 base pair (bp) DNA deletion found in the mitochondrial genome.

ALPHA GLOBIN GENE DNA MARKERS

Standard DNA mapping by Southern blotting was undertaken to characterise DNA polymorphisms and rearrangements at this locus (Hertzberg, Mickleson and Trent 1988). Over 400 haplotypes were established for Polynesians including Samoans, Maoris, Niueans and Tongans. Haplotypes were initially obtained from family studies and homozygous individuals and were then derived for those who were heterozygous for DNA polymorphisms.

Six haplotypes (Ia, IIa, IId, IIe, IIIa, IVa) predominated. A novel haplotype (IIIh), not described previously, was identified in Samoans (Table 1).

Polynesians and Melanesians share in common haplotypes IIIa and IVa. On the other hand, haplotypes Ia, IIa, IId, and IIe were frequently found in Polynesians but were virtually absent in Melanesians. This suggested a genetic contribution to pre-Polynesians from an ancestral population whose origins lay further to the west of island Melanesia. Haplotype Ia is also common in Europeans (Higgs *et al.* 1986). However, its presence at such high frequencies in Polynesians is unlikely to represent European admixture, particularly in view of HLA analyses by Serjeantson *et al.* (1987) which confirmed a paucity of European admixture in the Polynesians studied.

Indo-Pacific Prehistory Assn. Bulletin 11, 1991:270-275 (P. Bellwood ed, *Indo-Pacific Prehistory 1990*, Vol 2)

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Haplotype	Samoan No.(freq.)	Maori No.(freq.)	Niuean No.(freq.)	Tongan No.(freq.)	TOTAL No.(freq.)
Ia	67(.29)	23(.23)	41(.73)	16(.44)	147(.35)
IIa	13(.06)	11(.11)	1(.02)	1(.03)	26(.06)
IId	35(.15)	6(.06)	3(.05)	2(.06)	46(.11)
IIe	31(.13)	5(.05)	1(.02)	6(.17)	43(.10)
IIIa	44(.19)	8(.08)	2(.04)	4(.11)	58(.14)
IIIh	5(.02)	0	0	1(.03)	6(.01)
IVa	33(.14)	40(.40)	6(.11)	2(.06)	81(.19)
Other	IIf(1),IIc(1), IIg(1),I(1),IIIb(1)	Ie(1),IVc(1), Vc(1),IIIb(1), Unclass.(2)	IIf(1),IV(1)	IIf(3),IIb(1)	17(.04)
Total	233	99	56	36	424

TABLE 1: TOTAL ALPHA GLOBIN GENE HAPLOTYPES FOR NORMAL ALLELES IN POLYNESIANS

BETA GLOBIN GENE DNA MARKERS

Beta globin gene haplotypes reflecting 80 chromosomes from Samoans were compared to haplotypes reported for Southern Chinese (Trent *et al.* 1990). Haplotypes 1, 2, and 6 made up 60-62% of the total in Polynesian Samoans and Southern Chinese (Table 2). Only two of 10 haplotypes present in Mediterraneans were found to any significant frequency in the Samoans or Chinese. The similarity of haplotype patterns in Samoans and Southern Chinese is consistent with a common ancestral origin.

PHENYLALANINE HYDROXYLASE (PAH) GENE LOCUS

The PAH gene represents one of the most polymorphic DNA markers in the human genome (Lidsky *et al.* 1985). To date, only 46 distinct RFLP (restriction fragment length polymorphism) haplotypes have been obtained, which is significantly less than the 384 possible combinations at this locus. In the present study, 8 RFLPs were utilised to construct PAH-specific DNA haplotypes (Hertzberg *et al.* 1988). A total of 346 Polynesians (Samoans, Tongans, Cook Islanders, Maoris, Niueans) were studied.

Haplotypes 4, 1, and 7 accounted for the vast majority of the PAH alleles in each of the above five Polynesian groups (Table 3). A similar distribution was found for a Southeast Asian control group with the striking feature being the predominance of haplotype 4 in all Polynesians (58%) as well as the Southern Chinese (72%). In Europeans, haplotype 4 has an average frequency of 19% (range 13-32%) (Daiger *et al.* 1989).

An important finding from PAH data is the paucity of genetic variation at this locus in Polynesians since only a few common haplotypes were observed. This is comparable with the limited number of alpha globin gene haplotypes detected. The restricted diversity of

haplotypes in Polynesians supports the theory of a common, relatively small founding population into Polynesia. A second observation is the similarity of haplotypes found in Polynesians and Southeast Asians.

Haplotype	% in Chinese	% in Samoans	% in Mediterraneans
1	8	10	x
2	32	27	10
3	7	0	x
4	0	0	x
5	0	2	x
6	20	25	18
7	1	0	x
8	4	0	3
9	2	1	x
10	2	0	x
11	2	0	x
12	2	1	x
13	1	0	x
14	1	0	7
15	nd	4	x
16	nd	2	x
17	nd	1	x
18	nd	1	x
19	nd	1	x
20	nd	1	x
21	nd	1	x
22	nd	5	34
23	nd	1	x
Undefined	17	17	-

TABLE 2: BETA GLOBIN GENE HAPLOTYPE DISTRIBUTION AND COMPARISONS IN SAMOANS, CHINESE AND MEDITERRANEANS

Chinese data from Chan *et al.* 1986. nd = not described. Mediterranean data from Kazazian *et al.* 1984. x = comparable haplotypes not described.

MITOCHONDRIAL (MT) DNA MARKERS

A type of evolutionary change in Mt DNA involves length mutations (Cann and Wilson 1983). Nine have been described in the regions designated I-IX. One of these is due to a

deletion of one copy of a 9 bp tandem repeat sequence (CCCCCTCTA) within the non-coding region V (Wrischnik *et al.* 1987). Analyses to date have suggested that the latter is a valuable anthropological marker for peoples of East Asian origin. In the present study, the 9 bp deletion was detected by use of the polymerase chain reaction (PCR) (Hertzberg *et al.* 1989).

Haplotype*	Samoan No.(freq)	Tongan No.(freq)	Cook Is. No.(freq)	Maori No.(freq)	Niuean No.(freq)	Total Polynesian	Total SE Asian
1	37(.20)	18(.24)	43(.20)	20(.26)	18(.25)	136(.22)	6(.08)
2	0	0	0	0	0	0	0
3	3(.02)	1(.01)	0	2(.03)	0	6(.01)	1(.01)
4	116(.62)	43(.58)	114(.53)	40(.51)	48(.65)	361(.58)	52(.72)
5	0	0	0	1	1	2(<.01)	0
6	1(.01)	0	4(.02)	0	0	5(.01)	0
7	24(.13)	10(.14)	50(.23)	13(.17)	5(.07)	102(.16)	7(.10)
20	3(.02)	0	0	0	0	3(<.01)	0
31	2(.01)	0	0	0	0	2(<.01)	0
44	0	0	0	0	0	0	5(.07)
Unclassified	2(.01)	2(.03)	3(.01)	2(.03)	0	9(.01)	1(.01)
TOTAL	188	74	214	78	72	626	72

TABLE 3: RFLP HAPLOTYPES AT THE PAH LOCUS IN POLYNESIANS AND SOUTHEAST ASIANS
Nomenclature from Chakraborty *et al.* 1987.

200 Polynesians were studied. Controls included DNA from 30 Papua New Guinea highlanders, 28 coastal New Guineans, 40 Tolais from New Britain, 52 Fijians and 124 Australian Aborigines.

The Asian-specific 9 bp deletion was found in 93% of Polynesians from five distinct island groups (Table 4). Lower, but significant, deletion frequencies of 8% and 14% were found in coastal and island Melanesians respectively. The 9 bp deletion was not found in PNG highlanders. Sporadic findings of this marker in Australian Aborigines were also associated with alpha globin gene deletions, which are Southeast Asian specific, and would suggest that the Mt DNA data reflected recent genetic admixture. 79% of Fijians were also positive for the 9 bp Mt DNA deletion (Table 4).

Polynesians have the highest reported incidence for the 9 bp Mt DNA deletion. This provides the most convincing evidence for a Southeast Asian origin for the Austronesian speaking pre-Polynesians. The smaller frequencies in coastal and island Melanesians indicate a degree of Austronesian genetic admixture as a consequence of the spread of the founding ancestral Polynesians across Melanesia. In contrast, the virtual absence of the Mt DNA deletion in PNG highlanders and Australian Aborigines reflects the antiquity and relative geographic isolation of these two populations. The fact that 79% of Fijians had the Mt DNA would argue for an initial pre-Polynesian colonization of Fiji.

Population	Sample No.	Mt DNA Deletion No. Detected	% Positive
Samoans	40	39	98
Maoris	40	40	100
Niueans	40	40	100
Cook Islanders	40	34	85
Tongans	40	33	93
Total Polynesians	200	168	93
PNG Highlanders	30	0	0
Coastal PNG (Madang)	28	4	14
Tolais	40	3	8
Fijians	52	41	79
Australian Aborigines	124	4	3.6

TABLE 4: REGION V 9-bp DELETION OF Mt DNA IN POLYNESIANS AND OTHER PACIFIC ISLAND POPULATIONS

CONCLUSIONS

Nuclear and Mt DNA data were sought in Polynesians, Southeast Asians and other Pacific Island groups. A variety of unrelated nuclear DNA markers were studied to reduce the potential effect of natural selection (e.g. thalassaemia - alpha and beta globin gene loci - and malaria). Mt DNA provides an even more useful tool for studying populations since there is considerable homogeneity within the one individual; inheritance is maternal and there is a rapid rate of evolution. Results obtained in all instances showed marked genetic similarity between the Polynesians and Southeast Asians. Relatively few of the markers were found in Polynesians, which is consistent with founder effects enhanced by genetic drift. That there must have been stringent bottleneck(s) in the number of colonizing females into Polynesia was reflected in the virtual fixation of the 9 bp Mt DNA marker.

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