Chen, Liang Zhong, ***Genetic Affinities of Oceanic Populations Based on RFLP and Haplotype Analysis of Genetic Loci on Three Chromosomes*** , Human Biology, 64:1 (1992:Feb.) p.1

Genetic Affinities of Oceanic Populations Based on RFLP and Haplotype Analysis of Genetic Loci on Three Chromosomes

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Abstract Restriction fragment length polymorphisms at the renin and factor 13B loci located at chromosome 1q32–1q42 were studied in 14 ethnic groups in the west Pacific region. The allele frequencies were combined with previously described β-globin and albumin–vitamin D binding protein haplotype frequencies and used to assess genetic affinities among eight major ethnic-geographic groups in this region. These population groups divide into two clusters with Australian Aborigines, Island Melanesians, and Highland Melanesians forming one cluster and east Asians, Southeast Asians, Micronesians, and Polynesians forming the other. The results indicate that Micronesians and Polynesians are derived from populations in Southeast Asia and that they originated independently of the Melanesian populations.

The great diversity of peoples living in the western Pacific region can be divided into the following broad groups: Melanesians, Polynesians, Micronesians, Australians, east Asians, and Southeast Asians. The affinities and evolutionary history of these population groups is of considerable interest. A central issue in the prehistory of Oceania has been the historical relationships between the peoples of Melanesia, Micronesia, and Polynesia and the neighboring populations in Asia and Australia. Relevant archeologic and linguistic data have been reviewed by Howells (1973) and Bellwood (1978, 1989). Much discussion about Oceanic prehistory has focused on the distribution of Lapita pottery (Green 1979). White et al. (1988) suggest that the development of language and other aspects of culture that gave rise to the Lapita tradition evolved within the Bismarck Archipelago in Melanesia; others, for example, Terrell (1986), believe that Polynesians stemmed from this localized develop-

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ment. However, recent blood genetic evidence (Serjeantson and Hill 1989; Hertzberg, Mickleson et al. 1989) supports the longer established view that Polynesians and Austronesian-speaking Melanesians represent a separate wave of migration from east Asia.

Here, we extend the genetic information available for selected populations in the Asian-Pacific region with a description of molecular data for the renin (REN) and factor 13B (F13B) genes. The REN gene spans 12 kb and contains 10 exons separated by 9 intervening sequences. Hardman et al. (1984) isolated and characterized two renin genomic clones in the recombinant phage λHRIII and λHRV, which represent overlapping regions of the gene. The REN gene was first localized to 1q21–qter by DNA analysis of somatic cell hybrids (Naylor et al. 1983; Chirgwin et al. 1984). McGill et al. (1987) and Cohen-Haguenauer et al. (1987) mapped the gene to 1q25–q32 and 1q32 by in situ hybridization. More recently, Nakai et al. (1988) localized the gene to band 1q42.

Coagulation factor 13 is a tetramer (A_2B_2) consisting of two A subunits and two B subunits. The A and B subunits are products of different autosomal genes (Board 1979, 1980). The B subunit gene (F13B) was located at chromosome position 1q31–32.1 by in situ hybridization (Webb et al. 1989). Genetic variants of both subunits were first identified by Board (1979, 1980), and population variation of F13B alleles 1, 2, and 3 has been described by Board (1980), Dykes and Polesky (1987), Nishigaki and Omoto (1982), Kuhnl et al. (1983), Liefheit et al. (1985), Mauff et al. (1986), Miller et al. (1985), Nakamura and Abe (1982), Olaissen et al. (1983), Kamboh and Ferrell (1986), and Webb et al. (1989). The cDNA for F13B contains 2180 base pairs coding for the mature protein of 641 amino acids (Ichinose et al. 1986). This has been used to identify restriction fragment length polymorphisms (RFLPs) of F13B, possibly caused by an insertion or deletion event (Webb et al. 1989). The recombination frequency between REN and F13B is reported to be 18.5% by O'Connell et al. (1989) and 10% by Griffiths et al. (1989).

In this article we report studies of RFLPs at the REN and F13B loci in Oceanic populations; linkage disequilibrium between the loci is analyzed and gene frequencies are estimated. The results are combined with data obtained for the same populations for the β-globin and albumin–vitamin D binding protein gene complexes (Chen et al. 1990; Chen, Easteal, Board, and Kirk 1990) to investigate population affinities. The results are assessed in the context of previous studies of the genetics of these population groups.

Populations and Methods

The locations of populations sampled are shown in Figure 1. The sample included four Melanesian groups (Highlanders from the Eastern

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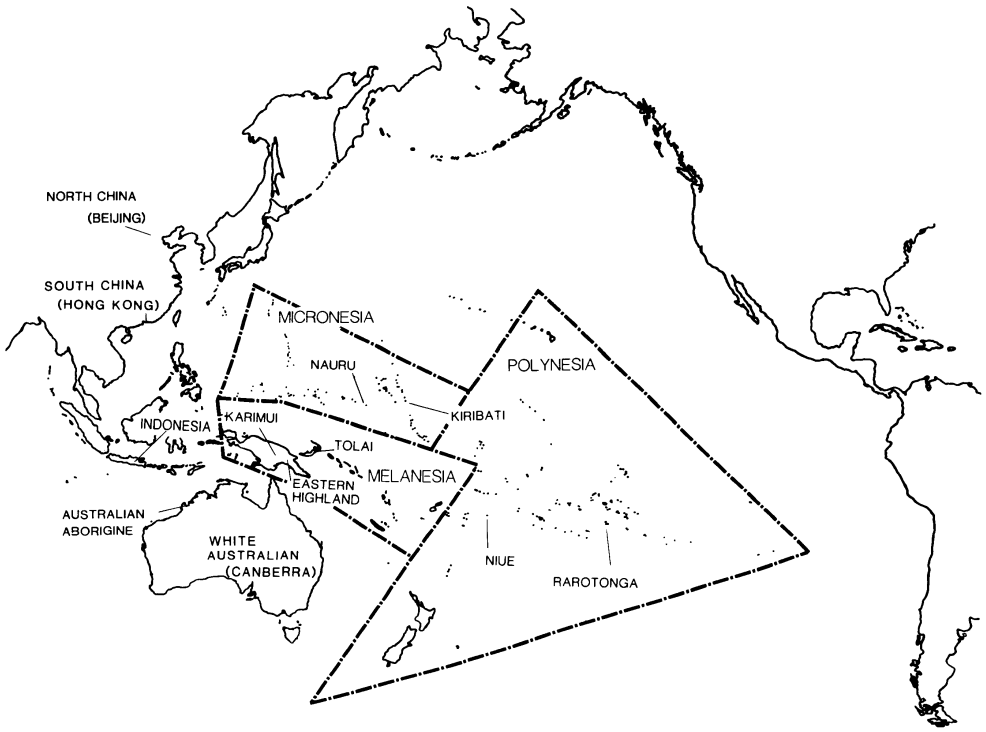

Figure 1. Locations of the sampled populations.

Highlands of Papua New Guinea, the Karimui from the fringe area of the Papua New Guinea highlands, Island Melanesians from Madang in the coastal area of Papua New Guinea, and Tolais on the island of New Britain), two Polynesian groups from the Cook Islands (Rarotonga) and Niue, two Micronesian groups from Nauru and Kiribati, two east Asian groups (northern Chinese from Beijing and southern Chinese from Hong Kong), one Southeast Asian group (the Javanese from Jogjakarta in Indonesia), two Australian aboriginal groups from northwest Australia, and white Australians from Canberra representing a European group. Wherever possible, 50 unrelated individuals (100 chromosomes) from each population were screened.

DNA samples were extracted from buffy coats, each sample (7 μg) was digested with 50 U Bgl II at 37°C overnight. The electrophoresis, blotting, hybridization, and autoradiography were carried out using standard methods (Maniatis et al. 1982; Grunebaum et al. 1984; Yenchitsomanus et al. 1985). When used as a hybridization probe, the genomic clone (λHRV) of the renin gene (Hardman et al. 1984) was found to hybridize with dispersed repeat elements and gave an unacceptable background smear on Southern blots. To eliminate repetitive elements from the probe, four genomic EcoR I fragments from λHRV were subcloned

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Figure 2. Southern blots of Bgl II RFLPs at both the REN and F13B loci; two probes are hybridized simultaneously on the same filter.

into pUC18. From 5′ to 3′, these 4 fragments are (1) pREN3k, which contains exons 2 and 3; (2) pREN2646, with exons 4 and 5; (3) pREN886, with exon 5A; and (4) pREN5k, with exons 6, 7, 8, and 9 (Chen and Easteal 1989). pREN886 was subsequently used as a probe because it did not hybridize to repetitive elements and because it appears to contain a unique sequence.

The cDNA F13B probe with 2.2-kb EcoR I insert (Ichinose et al. 1986) and pREN886 were labeled by nick translation (Rigby et al. 1977) to a specific activity at least 1×10^8 dpm/μg. Among the six enzymes (BamH I, Bgl II, EcoR I, HgiA I, Hinc II, and Hind III) tested, Bgl II digestion and hybridization with pREN886 was found to detect a single two-allele polymorphism at the REN locus, with variant bands of approximately 27 kb (allele 1) and 21 kb (allele 2). No invariant bands were observed.

Three enzymes, Bgl II, EcoR I, and Xba I, detect the same polymorphism at the F13B locus (Webb et al. 1989). In this study Bgl II was used because it reveals RFLPs for both REN and F13B. The hybridized bands are well separated from each other (27 kb and 21 kb for REN and 7.21 kb and 6.84 kb for F13B) so that both probes could be used at the same time to hybridize the RFLPs for both genes on the same filter (Figure 2).

Linkage disequilibrium was analyzed using log-linear modeling techniques (Weir and Wilson 1986) with the computer program GLIM (version 3.77). Patterns of relatedness among populations were assessed in three ways: (1) the unweighted pair-group method with arithmetic averages (UPGMA; Sneath and Sokal 1973) based on matrices of Nei's (1978) unbiased minimum genetic distance; (2) the Wagner distance parsimony

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Table 1. REN RFLP Genotype and Allele Frequencies

		Genotype Frequency			*Allele Frequency*		
Population	*Number Tested*	*27 kb* (REN 1)	*27/21 kb* (REN 1–2)	*21 kb* (REN 2)	*REN*1*	*REN*2*	*S.E.*
Melanesia							
Madangs	59	0	4	55	0.0339	0.9661	0.017
Tolais	50	0	7	43	0.0700	0.9300	0.026
Eastern Highlanders	71	0	2	69	0.0141	0.9859	0.010
Karimuis	65	0	10	55	0.0769	0.9231	0.023
Polynesia							
Niueans	50	0	7	43	0.0700	0.9300	0.026
Cook Islanders	46	1	5	40	0.0761	0.9239	0.028
Mixed	20	0	4	16	0.1000	0.9000	0.047
Micronesia							
Kiribats	46	0	5	41	0.0543	0.9457	0.024
Nauruans	53	3	20	30	0.2453	0.7547	0.042
Southeast Asia							
Javanese	52	0	7	45	0.0673	0.9327	0.025
East Asia							
Northern Chinese	48	3	8	37	0.1458	0.8542	0.036
Southern Chinese	40	2	10	28	0.1750	0.8250	0.042
Australia							
Aborigines	125	0	14	111	0.0560	0.9440	0.015
Europeans	56	2	19	35	0.2054	0.7946	0.038

method (Farris 1972) based on Cavalli-Sforza and Edwards's (1967) chord distance; and (3) partial maximum likelihood estimation (Felsenstein 1973a,b). The first two analyses were performed using the BIOSYS computer package (release 1, 1981; D.L. Swofford and R.B. Selander, University of Illinois, Urbana). For the maximum likelihood analysis the CONTML program from the PHYLIP computer package (version 3.0; J. Felsentein, University of Washington, Seattle) was used.

Results

Allele Frequence Distributions. The RFLPs of both REN and F13B show highly variable polymorphic frequencies in the Oceanic populations.

REN was screened in 781 individuals (Table 1). The allele frequency of *REN*1* is generally low in the Island and Highland Melanesians (0.04–0.05), Aborigines (0.06), Indonesians (0.07), and Polynesians (0.08). The Chinese and Micronesians have higher frequencies (0.16), and the white Australians rank the highest (0.21).

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Table 2. F13B RFLP Genotype and Allele Frequencies

		Genotype Frequency			*Allele Frequency*		
Population	*Number Tested*	*6.8 kb* (F13B a)	*6.8/7.2 kb* (F13B ab)	*7.2 kb* (F13B b)	*F13B*A*	*F13B*B*	*S.E.*
Melanesia							
Madangs	59	14	22	23	0.4237	0.5763	0.045
Tolais	50	8	14	28	0.3000	0.7000	0.046
Eastern Highlanders	66	18	27	21	0.4773	0.5227	0.043
Karimuis	65	26	27	12	0.6077	0.3923	0.043
Polynesia							
Niueans	30	0	8	22	0.1333	0.8667	0.044
Cook Islanders	46	1	6	39	0.0870	0.9130	0.029
Micronesia							
Kiribats	32	0	6	26	0.0937	0.9063	0.036
Nauruans	53	3	18	32	0.2264	0.7736	0.041
Southeast Asia							
Javanese	52	2	8	42	0.1154	0.8846	0.031
East Asia							
Northern Chinese	48	3	17	27	0.2447	0.7553	0.044
Southern Chinese	40	4	11	25	0.2375	0.7625	0.048
Australia							
Aborigines	89	10	43	36	0.3146	0.6854	0.035
Europeans	56	18	26	12	0.5536	0.4464	0.047

F13B was screened in 705 samples (Table 2). *F13B*A* has intermediate frequencies in the Melanesians, ranging from 0.30 in Tolais to 0.61 in Karimuis, and lower frequencies in the Polynesians and Micronesians, ranging from 0.09 in Rarotonga and Kiribati to 0.13 in Niue and 0.23 in Nauru. Indonesians show a frequency (0.12) similar to the frequencies in Polynesians and Micronesians. The two Chinese groups have frequencies of 0.24 and 0.26; the frequency in Aborigines (0.32) is close to Island Melanesians. The frequency in white Australians (0.55) is relatively high.

In none of the populations is the phenotypic distribution at either locus significantly different from Hardy-Weinberg expectations.

Linkage Disequilibrium. No significant linkage disequilibrium was observed between REN and F13B alleles except in the Rarotonga sample in which *REN*1* and *F13B*A* associate nonrandomly ($p < 0.05$) (Table 3). A single significant association in 14 comparisons can be expected to occur by chance. Thus there appears to be no linkage disequilibrium between these two loci. The apparent absence of linkage disequilibrium may reflect the distance between the loci or the loss or creation of restriction sites on multiple occasions.

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Table 3. Likelihood Criteria for Linkage Disequilibrium between REN and F13B

Population	*Number Tested*	*Scaled Deviance*	*p (d.f. = 4)*
Melanesia			
Madangs	59	2.2584	>0.50
Tolais	50	1.3277	>0.80
Eastern Highlanders	71	1.3301	>0.80
Karimuis	65	0.8193	>0.80
Polynesia			
Niueans	50	0.6334	>0.90
Cook Islanders	46	11.159	<0.05
Micronesia			
Kiribats	46	1.3162	>0.80
Nauruans	53	0.4367	>0.90
Southeast Asia			
Javanese	52	3.8648	>0.30
East Asia			
Northern Chinese	48	5.5309	>0.20
Southern Chinese	40	3.0807	>0.50
Australia			
Kalumburu	30	2.5426	>0.50
Mowanjum	95	3.9382	>0.30
Europeans	56	4.5570	>0.20

Population Affinities. The REN and F13B allele frequencies are combined with the previously published β-globin (Chen, Easteal, Board, and Kirk 1990) and ALB-GC-DBP (Chen et al. 1990) haplotype frequencies to obtain matrices of Nei's (1978) unbiased minimum distance and Cavalli-Sforza and Edwards's (1967) chord distance (Table 4) for the following ethnic and regional population groups: white Australians,

Table 4. Genetic Distance Coefficients (×1000) Based on the Pooled Data of Haplotypes and RFLPs for the REN, F13B, β-globin, and ALB-GC-DBP Loci

Population	*1*	*2*	*3*	*4*	*5*	*6*	*7*	*8*
1 Island Melanesians	–	21	42	37	13	27	49	39
2 Highland Melanesians	252	–	62	43	35	28	57	30
3 Polynesians	262	244	–	6	50	13	6	101
4 Micronesians	291	234	167	–	46	6	8	76
5 Aborigines	278	246	281	314	–	36	54	50
6 East Asians	272	189	162	180	286	–	16	50
7 Southeast Asians	310	244	176	195	312	222	–	99
8 White Australians	319	273	343	341	368	260	368	–

Below diagonal: Cavalli-Sforza and Edwards (1967) chord distance. Above diagonal: Nei's (1978) unbiased minimum distance.

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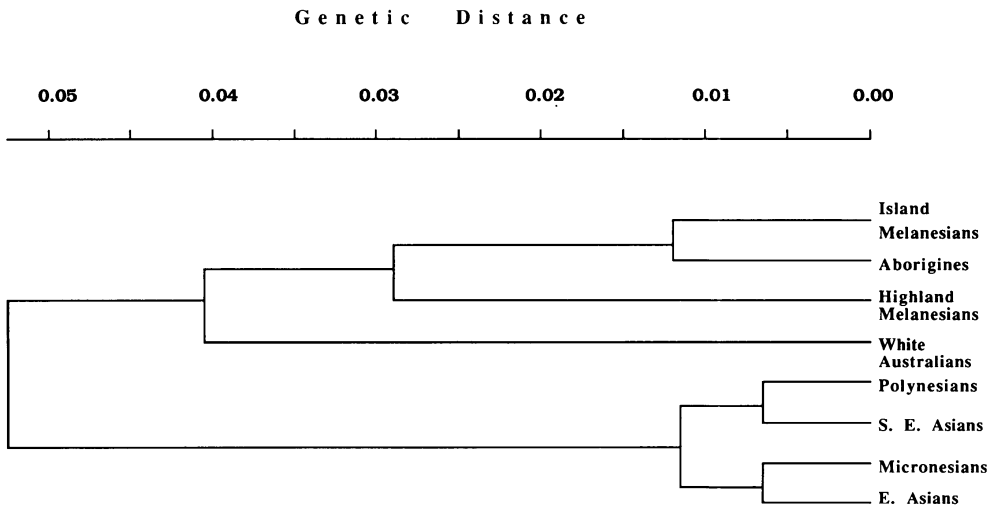

Figure 3. UPGMA phenogram of population groups based on Nei's unbiased minimum distance.

Australian Aborigines, Highland Melanesians, Island Melanesians, Polynesians, Micronesians, Southeast Asians, and east Asians. A UPGMA phenogram (Figure 3) and a Wagner parsimony tree with midpoint rooting (Figure 4) were constructed from Nei and Cavalli-Sforza and Edwards's distances, respectively. A network was also obtained by partial maximum likelihood estimation. The log-likelihood value of this network was compared with that of 11 other networks. It was not significantly different from any of them (Figure 5).

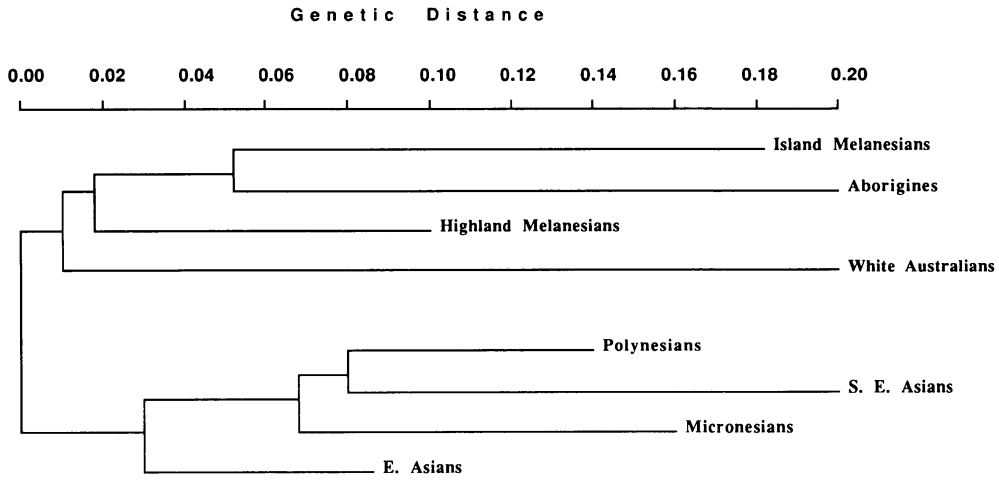

Figure 4. Wagner parsimony tree of populations groups based on Cavalli-Sforza and Edwards's chord distance with midpoint rooting.

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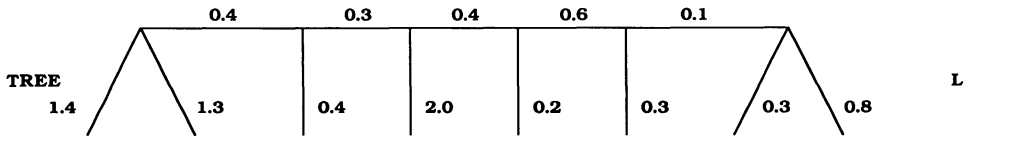

1	{[(AB,	IM)	HM]	[EU	[EA	[MI	(PO,	ID)]]]}	327.4
2	{[(AB,	IM)	HM]	[EU	[EA	[(MI,	PO)	ID]]]}	327.1
3	{[(AB,	IM)	HM]	[EU	[EA	[(MI,	ID)	PO]]]}	327.1
4	{[AB	(IM,	HM)]	[EU	[EA	[MI	(PO,	ID)]]]}	326.8
5	{[(AB,	HM)	IM]	[EU	[EA	[MI	(PO,	ID)]]]}	326.8
6	{[(AB,	IM)	HM]	[EU	[(EA,	ID)	(MI,	PO)]]}	323.3
7	{[(AB,	IM)	HM]	[EU	[ID	[EA	(MI,	PO)]]]}	323.3
8	{[(AB,	IM)	HM]	[EU	[(EA,	PO)	(MI,	ID)]]}	320.0
9	{ (AB,	HM)	[EU	[EA	[IM	[MI	(PO,	ID)]]]]}	324.0
10	{ (AB,	HM)	[EU	[EA	[ID	[IM	(PO,	MI)]]]]}	322.7
11	{[[(AB,	IM)	HM]	ID]	[EU	[EA	(PO,	MI)]]}	320.0
12	{[(AB,	HM)	(ID,	IM)]	[EU	[EA	(PO,	MI)]]}	319.2

Figure 5. Log-likelihood results (L) for 12 networks joining the 8 population groups. The maximum likelihood network (1) is shown with relative branch lengths ($\times 10^{-2}$) indicated. AB, Australian Aborigines; IM, Island Melanesians; HM, Highland Melanesians; EU European Australians; EA, East Asians (Chinese); MI, Micronesians; PO, Polynesians; ID, Indonesians.

The three tree building methods give reasonably consistent results. If the white Australians are taken as an "out group," then the other population groups form two distinct clusters: Australian Aborigines and Melanesians in one group and the remaining populations in the other. In the first group Aborigines consistently appear most closely related to the Island Melanesians. In the second group the UPGMA phenogram indicates a close affinity between Micronesians and east Asians, whereas the other two methods indicate a clustering of Micronesians, Polynesians, and Southeast Asians separate from east Asians.

Discussion

The results obtained here are broadly consistent with a number of previous studies of the genetics of these population groups. Summarizing the protein genetic markers of four highly polymorphic loci (proteinase inhibitor, transferrin, group-specific component, and phosphoglucomutase-1), Kamboh (1984) showed that Australian Aborigines from the central desert area and Papua New Guineans from the Eastern Highlands and Sepik River area have close affinities with each other, and they form a branch in a phenogram based on Nei's genetic distance that is the earliest to split. The other Asians and Pacific populations form a separate group.

One of the interesting findings in this study is the striking similarity between the series from Australian Aborigines (northwest Australia) and Melanesians from Papua New Guinea, especially the Tolais from Rabaul, New Britain. This affinity is also indicated by the separate analysis

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of ALB-GC-DBP haplotypes (Chen, Easteal, Board, and Kirk 1990) but not of the β-globin haplotypes (Chen et al. 1990).

Previous studies based on other blood genetic markers have noted the close relationships between Aborigines and Papua New Guinea Highlanders [e.g., Kamboh (1984), Serjeantson et al. (1982), Keats (1977), and Schanfield (1977)], and Keats (1977) showed that the Anga, an intermediate Papuan population, were indeed very close to Aborigines, particularly those from central Australia. Unfortunately, there is little previous blood genetic information from the Tolais, so direct comparison with the present results is not possible. However, it is worth noting that Howells (1973), using multivariate techniques utilizing data on skull measurements, reported a close relationship between the Aborigines and the Tolais, although this result has not been substantiated by other workers using similar techniques (Giles 1976).

The genetic relationship between the Aborigines and the Tolais and between the Karimuis and Papua New Guinea Highlanders is consistent with what is known of the prehistory of the region. The first movement of people into Sahul-land (i.e., the single Pleistocene landmass of Australia and New Guinea) took place 50,000 years ago (Bellwood 1989). Wurm (1983) suggested on the basis of linguistic evidence that a second migration into the area was by Papuan-speakers and that this occurred before the end of the Pleistocene Epoch. Recent excavations have provided evidence for occupation on New Ireland at least 30,000 years ago (White et al. 1988). Thus it is plausible to suggest that groups of genetically related populations were widespread across Sahul-land and reached nearby islands 30,000–40,000 years ago. Although subsequent migrations and population movements have modified the original genetic background of their populations, there are sufficient similarities remaining for this group of Papua New Guinea and Australian aboriginal populations to be genetically close, as indicated by the present analysis of DNA RFLPs and haplotypes.

In the present investigation and in the individual analyses of the β-globin and albumin–vitamin D binding protein haplotypes (Chen, Eastreal, Board, and Kirk 1990; Chen et al. 1990), the two populations representing Polynesians (Niue, Rarotonga) and the two representing Micronesians (Kiribati and Nauru) nearly always cluster together and are separate from the Melanesian and Australian aboriginal populations.

These results support the view that Polynesians are genetically distinct from Melanesians and are not derived by local evolution from a former Melanesian group. The Polynesians and the Micronesians are most closely allied genetically with east and Southeast Asians, a view that corresponds with that based on linguistic evidence, which places the origin of the Austronesian language in the area of Taiwan.

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The genetic affinity of the Polynesians with the Southeast Asians and their distinction from the Melanesians detected in the present study is supported by a number of studies involving a range of distinct genetic loci. In their study of HLA types in the Pacific, Serjeantson et al. (1987) concluded that Polynesians are genetically distinct from Melanesians and share some HLA types with Southeast Asians. The detailed studies of the α-globin gene complex in Polynesians by Hertzberg et al. (1988) have also revealed two haplotypes that enable Polynesians to be distinguished from Melanesians, and Hertzberg concluded that three of the haplotypes present in Polynesians suggest "a genetic component derived from an ancestral population, whose origin may lie to the west of Island Melanesia" (p. 975). The analysis of RFLP haplotypes at the phenylalanine hydroxylase (PAH) locus in Polynesians also points to an evolutionary origin in Southeast Asia. Hertzberg, Jahromi et al. (1989) showed that the three most common PAH haplotypes in Polynesia are shared with a diverse Southeast Asian group.

Some of the most compelling evidence for an evolutionary relationship between Southeast Asians and Polynesians has been obtained from the study of mitochondrial DNA (mtDNA). Wrischnik et al. (1987) suggested that a particular 9-bp deletion in the mt genome is an anthropologic marker for people of Southeast Asian origin. Hertzberg, Mickleson et al. (1989) showed that this deletion reaches very high frequencies in Polynesians (93%). In contrast, this deletion is found only occasionally in coastal Melanesians from Papua New Guinea and is absent from Papua New Guinea Highlanders. In addition, the deletion was detected in only one Australian Aborigine, who also had a typical Southeast Asian ζ-globin gene rearrangement, indicating some Southeast Asian ancestry.

Notwithstanding the genetic evidence pointing to a Southeast Asian evolutionary origin for the Polynesians, there is also evidence suggesting a distinct Melanesian contribution to the Polynesian gene pool. For example, the $-\alpha^{3.7}$ III thalassemia mutation that occurs in Island Melanesians is also found in Polynesians (Hill et al. 1985, 1987; Trent et al. 1988). Furthermore, Hertzberg et al. (1988) showed that the $-\alpha^{3.7}$ III deletion occurs on the same RFLP haplotype in Melanesians and Polynesians, confirming the suggestion of Hill et al. (1985) that the deletion occurred only once. However, because the $-\alpha^{3.7}$ III deletion is only rarely detected in Papua New Guinea, Yenchitsomanus et al. (1985) suggested that it may have arisen in eastern Melanesia or Polynesia and diffused west.

Thus, taken together, the available genetic data support the widely held view [reviewed by Bellwood (1978) and Serjeantson and Hill (1989)] that the Pacific was colonized by successive waves of migration. The Papua New Guinea Highlanders and Australian aboriginal populations

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represent the early Australoid migration, which was superseded in coastal Papua New Guinea and Island Melanesia by later migrations from east Asia that ultimately populated Micronesia and Polynesia.

The results obtained by the three different methods of phylogenetic reconstruction are broadly consistent with each other. The consistency of these results with those found in previous genetic studies supports the validity of the general pattern of relationship indicated in Figures 3–5. However, the results are based on too few data to allow firm acceptance of this pattern. This is demonstrated by the lack of significant difference between the maximum likelihood network and the 11 other networks shown in Figure 5, which includes 2 networks (9 and 10) in which Island Melanesians are clustered with Indonesians, Polynesians, and Micronesians. The pattern of relationship presented here is the best estimate for the available data. The allelic distribution of many more genes will need to be determined before a pattern can be established definitively.

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Literature Cited

Bellwood, P.S. 1978. *Man's Conquest of the Pacific*. New York: Oxford University Press.

Bellwood, P.S. 1989. The colonization of the Pacific: Some current hypotheses. In *The Colonization of the Pacific: A Genetic Trail,* A.V.S. Hill and S.W. Serjeantson, eds. Oxford: Oxford University Press, 1–59.

Board, P.G. 1979. Genetic polymorphism of the A subunit of human coagulation factor XIII. *Am. J. Hum. Genet.* 31:116–124.

Board, P.G. 1980. Genetic polymorphism of the B subunit of human coagulation factor XIII. *Am. J. Hum. Genet.* 32:348–353.

Cavalli-Sforza, L.L., and A.W.F. Edwards. 1967. Phylogenetic analysis: Models and estimation procedures. *Evolution* 21:550–570.

Chen, L.Z., and S. Easteal. 1989. Renin locus restriction fragment length polymorphism. *Hum. Genet.* 82:302.

Chen, L.Z., S. Easteal, P.G. Board, and R.L. Kirk. 1990. Evolution of β-globin haplotypes in human populations. *Mol. Biol. Evol.* 7:423–437.

Chen, L.Z., S. Easteal, P.G. Board, K.M. Summers, K.K. Bhatia, and R.L. Kirk. 1990. Albumin–vitamin D binding protein haplotypes in Asian-Pacific populations. *Hum. Genet.* 85:89–97.

Chirgwin, J.M., I.M. Schaefer, P.S. Rotwein, N. Piccini, K.W. Gross, and S.L. Naylor. 1984. Human renin gene is on chromosome 1. *Somat. Cell Mol. Genet.* 10:415–421.

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Chen, Liang Zhong, ***Genetic Affinities of Oceanic Populations Based on RFLP and Haplotype Analysis of Genetic Loci on Three Chromosomes*** , Human Biology, 64:1 (1992:Feb.) p.1

Cohen-Haguenauer, O., F. Soubrier, V. Nguyen, M.S. Gross, C. Jegou, P. Corvol, and J. Frezal. 1987. Confirmation of the chromosomal assignment of the renin gene to human chromosome 1 and regional assignment using in situ hybridization. *Cytogenet. Cell Genet.* 46:597.

Dykes, D.D., and H.F. Polesky. 1987. The use of the serum protein factor XIIIB (FXIIIB) in parentage testing. *Am. J. Clin. Pathol.* 88:627–630.

Farris, J.S. 1972. Estimating phylogenetic trees from distance matrices. *Am. Natur.* 106:645–668.

Felsenstein, J. 1973a. Maximum likelihood and minimum-step methods for estimating evolutionary trees from data on discrete characters. *Syst. Zool.* 22:240–249.

Felsenstein, J. 1973b. Maximum-likelihood estimation of evolutionary trees from continuous characters. *Am. J. Hum. Genet.* 25:471–492.

Giles, E. 1976. Cranial variation in Australia and neighbouring areas. In *The Origin of the Australians,* R.L. Kirk and A.G. Thorne, eds. Canberra: Australian Institute of Aboriginal Studies, 161–172.

Green, R.C. 1979. Lapita. In *The Prehistory of Polynesia,* J.D. Jennings, ed. Cambridge, MA: Harvard University Press, 27–60.

Griffiths, L.R., P.G. Board, M.B. Zwi, B.J. Morris, J.G. McLeod, and G.A. Nicholson. 1989. The B subunit of coagulation factor XIII is linked to renin and the Duffy blood group to α-spectrin on human chromosome 1. *Hum. Hered.* 39:107–109.

Grunebaum, L., J.P. Cazenave, G. Camerino, C. Kloepfer, J.L. Mandel, P. Tolstoshev, M. Jaye, H. de la Shalle, and J.P. Lecocz. 1984. Carrier detection of hemophilia B by using a restriction site polymorphism associated with the coagulation factor IX gene. *J. Clin. Invest.* 73:1491–1495.

Hardman, J.A., Y.J. Hort, D.F. Catanzaro, J.T. Tellam, J.D. Baxter, B.J. Morris, and J. Shine. 1984. Primary structure of the human renin gene. *DNA* 6:457–468.

Hertzberg, M.S., K.N.P. Mickleson, and R.J. Trent. 1988. α-globin gene haplotypes in Polynesians: Their relationships to population groups and gene rearrangements. *Am. J. Hum. Genet.* 43:971–977.

Hertzberg, M., K.N.P. Mickleson, S.W. Serjeantson, J.F. Prior, and R.J. Trent. 1989. An Asian-specific 9-bp deletion of mitochondrial DNA is frequently found in Polynesians. *Am. J. Hum. Genet.* 44:504–510.

Hertzberg, M.S., K. Jahromi, V. Ferguson, H.H.M. Dahl, J. Mercer, K.N.P. Mickleson, and R.J. Trent. 1989. Phenylalanine hydroxylase gene haplotypes in Polynesians: Evolutionary origins and absence of alleles associated with severe phenylketonuria. *Am. J. Hum. Genet.* 44:382–387.

Hill, A.V.S., B. Gentile, J.M. Bonnardot, J. Roux, D.J. Weatherall, and J.B. Clegg. 1987. Polynesian origins and affinities: Globin gene variants in eastern Polynesia. *Am. J. Hum. Genet.* 40:453–463.

Hill, A.V.S., D.K. Bowden, F.J. Trent, D.R. Higgs, S.J. Oppenheimer, S.L. Thein, K.N.P. Mickleson, D.J. Weatherall, and J.B. Clegg. 1985. Melanesian and Polynesians share a unique α-thalassemia mutation. *Am. J. Hum. Genet.* 37:571–580.

Howells, W.W. 1973. *The Pacific Islanders.* New York: Weidenfeld and Nicolson.

Ichinose, A., B.A. McMullen, K. Fujikawa, and E.W. Davie. 1986. Amino acid sequence of the *B* subunit of human factor XIII, a protein composed of ten repetitive segments. *Biochemistry* 25:4633–4938.

Kamboh, M.I. 1984. Population genetic studies using isoelectric focusing in the Asian, Pacific Australian area. Ph.D. thesis, Australian National University, Canberra, Australia.

Kamboh, M.I., and R.E. Ferrell. 1986. Genetic studies of low abundance human plasma proteins. II. Population genetics of coagulation factor XIIIB. *Am. J. Hum. Genet.* 39:817–825.

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Chen, Liang Zhong, ***Genetic Affinities of Oceanic Populations Based on RFLP and Haplotype Analysis of Genetic Loci on Three Chromosomes*** , Human Biology, 64:1 (1992:Feb.) p.1

Keats, B. 1977. Genetic structure of the indigenous populations in Australia and New Guinea. *J. Hum. Evol.* 6:319–339.

Kirk, R.L. 1980. Language, genes, and people in the Pacific. In *Population Structure and Genetic Disorders,* A.W. Eriksson, ed. London: Academic Press, 113–137.

Kuhnl, P., P. Krechel, and W. Spielmann. 1983. Coagulation factor XIIIB (FXIIIB) allotyping by immunofixation electrophoresis: Confirmation of the three-allele-model. *Electrophoresis* 82:767–776.

Leifheit, H.-J., A.G. Gathof, and S. Weidinge. 1985. Classification of factor XIII B allotypes by isoelectric focusing. *Artzl. Lab.* 31:159–163.

Maniatis, T., E.F. Fritsch, and J. Sambrook. 1982. *Molecular Cloning: A Laboratory Manual.* Cold Spring Harbor, NY: Cold Spring Harbor Laboratory.

Mauff, G., A. Schilling, and G. Pulverer. 1986. Coagulation factor XIII. Determination of the genetic polymorphism of subunit B (F13B) by immunoblotting in a West German population sample. In *Advances in Forensic Haemogenetics,* B. Brinkmann and K. Henningsen, eds. Berlin: Springer-Verlag, vol. 1, 453–457.

McGill, J.R., J.M. Chirgwin, C.M. Moore, and J.L. McCombs. 1987. Chromosomal localization of the human renin gene (REN) by in situ hybridization. *Cytogenet. Cell Genet.* 45:55–57.

Miller, S.A., D.D. Dykes, and H.F. Polesky. 1985. Gene frequency distribution of the B subunit of Factor XIII (FXIIIB) in Minnesota whites, blacks and Amerindians. *Electrophoresis* 6:399–401.

Nakai, H., S. Inoue, H. Miyazaki, K. Murakami, and K. Tada. 1988. Human renin gene assigned to chromosome band 1q42 by in situ hybridization. *Cytogenet. Cell Genet.* 47:90–91.

Nakamura, S., and K. Abe. 1982. Genetic polymorphism of coagulation factor XIII B-subunit. *Ann. Hum. Genet.* 46:203–207.

Naylor, S.L., I. Schaefer, P. Rotwein, N. Piccini, K. Gross, and J.M. Chirgwin. 1983. Human renin gene is located on chromosome 1. *Cytogenet. Cell Genet.* 3:549–550.

Nei, M. 1978. Estimation of average heterozygosity and genetic distance from a small number of individuals. *Genetics* 89:583–590.

Nishigaki, T., and K. Omoto. 1982. Genetic polymorphism of the B subunit of human coagulation factor XIII. *Jpn. J. Hum. Genet.* 27:265–270.

O'Connell, P., G.M. Lathrop, Y. Nakamura, M.L. Leppert, R.H. Ardinger, J.L. Murray, J.-M. Lalouel, and R. White. 1989. Twenty-eight loci from a continuous linkage map of markers for human chromosome 1. *Genomics* 4:12–20.

Olaissen, B., A. Siverts, T. Gedde-Dahl, Jr., R. Jonassen, and P. Teisberg. 1983. Coagulation factor 13A and B-polymorphisms. In *Advances in Forensic Haemogenetics,* B. Brinkmann and K. Henningsen, eds. Berlin: Springer-Verlag, vol. 1, 459–465.

O'Shaughnessy, D.F., A.V.S. Hill, D.K. Bowden, D.J. Weatherall, and J.B. Clegg. 1990. Globin genes in Micronesia: Origins and affinities of Pacific Island peoples. *Am. J. Hum. Genet.* 46:144–155.

Rigby, P.W.J., M. Dieckmann, C. Rhodes, and P. Berg. 1977. Labelling deoxyribonucleic acid to high specific activity in vitro by nick translation with DNA polymerase I. *J. Mol. Biol.* 113:237–251.

Schanfield, M.S. 1977. Population affinities of the Australian Aborigines as reflected by the genetic markers of immunoglobulins. *J. Hum. Evol.* 6:341–352.

Sealey, P.G., P.A. Whittaker, and E.M. Southern. 1985. Removal of repeated sequences from hybridization probes. *Nucleic Acids Res.* 13:1905–1922.

Serjeantson, S.W., and A.V.S. Hill. 1989. The colonization of the Pacific: The genetic evidence. In *The Colonization of the Pacific: A Genetic Trail,* A.V.S. Hill and S.W. Serjeantson, eds. Oxford: Oxford University Press, 286–294.

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Chen, Liang Zhong, ***Genetic Affinities of Oceanic Populations Based on RFLP and Haplotype Analysis of Genetic Loci on Three Chromosomes*** , Human Biology, 64:1 (1992:Feb.) p.1

Serjeantson, S.W., D.P. Ryan, and A.R. Thompson. 1982. The colonization of the Pacific: The story according to human leucocyte antigens. *Am. J. Hum. Genet.* 34:904–918.

Serjeantson, S.W., B.S. White, E.C. Jazwinska, P.T. Yenchitsomanus, K.N.P. Mickleson, and R.J. Trent. 1987. HLA-DR and -DQ DNA polymorphisms: New linkage relationships established by RFLP genomic typing in Polynesians and Melanesians. *Hum. Immunol.* 20:145–153.

Sneath, P.H.A., and R.R. Sokal. 1973. *Numerical Taxonomy: The Principles and Practice of Numerical Classification.* San Francisco, CA: Freeman, 230–234.

Sofro, A.S.M. 1983. Population genetic studies in Indonesia. Ph.D. thesis, Australian National University, Canberra, Australia.

Terrell, J. 1986. *Prehistory in the Pacific Islands.* Cambridge: Cambridge University Press.

Trent, R.J., J.B. Buchanan, A. Webb, R.P.S. Goundar, L.M. Seruvatu, and K.N.P. Mickleson. 1988. Globin genes are useful markers to identify genetic similarities between Fijians and Pacific Islanders from Polynesia and Melanesia. *Am. J. Hum. Genet.* 42:601–607.

Webb, G.C., M. Coggan, A. Ichinose, and P.G. Board. 1989. Localization of the coagulation factor XIIIB subunit gene (F13B) to chromosome bands 1q31–32.1 and restriction fragment length polymorphism at the locus. *Hum. Genet.* 81:157–160.

Weir, B.S., and S.R. Wilson. 1986. Log-linear models for linked loci. *Biometrics* 42:665–670.

White, P., J. Allen, and J. Specht. 1988. Peopling the Pacific: The Lapita Homeland Project. *Aust. Natur. Hist.* 22:410–416.

Wrischnik, L.A., R.G. Higuchi, M. Stoneking, H.A. Erlich, N. Arnheim, and A.C. Wilson. 1987. Length mutations in human mitochondrial DNA: Direct sequencing of enzymatically amplified DNA. *Nucleic Acids Res.* 15:529–542.

Wurm, S.A. 1983. Linguistic prehistory in the New Guinea area. *J. Hum. Biol.* 12:25–36.

Yenchitsomanus, P., K.M. Summers, K.K. Bhatia, J. Cattani, and P.G. Board. 1985. Extremely high frequencies of α-globin gene deletion in Madang and on Kar Kar Island, Papua New Guinea. *Am. J. Hum. Genet.* 37:778–784.

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