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An Ancient Common Origin of Aboriginal Australians and New Guinea Highlanders Is Supported by α-Globin Haplotype Analysis

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Summary

The origins of aboriginal Australians and their relationship with New Guineans and neighboring Southeast Asians remain controversial. We have studied the α-globin haplotype composition of an aboriginal tribe from central Australia, to address some of the ambiguities of previous studies. Australians have a haplotype repertoire that is shared with New Guinea highlanders, a fact that strongly supports a common origin of these two populations. Further, Australians and New Guinea highlanders have a different set of α haplotypes from Southeast Asians and a lower genetic diversity. This, coupled with the presence of many locally specific central Australian haplotypes, suggests that much of the original diversity was lost in a population bottleneck prior to or during the early colonization of Sahul and that subsequent recovery of diversity has been accompanied by the generation of new haplotypes. These conclusions contrast with some previous genetic studies suggesting links between Australians, coastal New Guineans, and present-day Southeast Asians. Much of this discrepancy appears to be due to more recent Southeast Asian admixture on the north coast of Australia.

Introduction

Australia was first settled ≥40,000 years ago from Southeast Asia (Roberts et al. 1990; Bowdler 1993) in a migration through the islands in the biogeographical area of Wallacea to the old continent of Sahul, which comprised Australia and New Guinea (fig. 1) (White and O'Connell 1982; Groube et al. 1986; Flood 1989, pp. 29–74). At times of lowered sea levels due to glaciation, the islands of Java, Borneo, and Bali were joined with Vietnam and the Malayan peninsula to form the large continent of Sunda. For any expanding or displaced Asian population of modern humans with seafaring abilities, Sahul was accessible throughout the late Pleistocene, requiring, on occasion, a sea voyage of perhaps only 70 km. The archaeological record suggests that migration also continued eastward of New Guinea, resulting in the occupation of the Bismarck archipelago and the northern Solomon Islands by 30,000 years ago (Bellwood 1978; Gosden 1993). However, questions remain about the number and nature of ancient source populations from Sunda (Flood 1989, pp. 29–74; Thorne and Wolpoff 1992; Bowdler 1993) and about the relationships between the peoples of the New Guinea highlands, who are thought to be direct descendants of the early colonists, and aboriginal Australians.

By 7,000 years ago, New Guinea and Australia had become islands permanently separated by the Torres Strait. Subsequent to this event, waves of Austronesian-speaking migrants began entering the old Sahul region from Southeast Asia. The Indo-Malaysian archipelago was settled perhaps as long as 5,000 years ago. Following this, an eastward expansion, which ultimately took the first colonizers to Polynesia, introduced distinctive genetic and linguistic influences to coastal New Guinea and island Melanesia. There was also a southeastward Melanesian expansion to the islands of the Southern Solomons, Vanuatu, and New Caledonia. Significantly, there are no traces in the archaeological record of any major migrations into Australia or the New Guinea highlands during this period (Bellwood 1978, 1985, 1989; Terrell 1986).

The archaeological, anthropological, and linguistic assessments of population migration and evolutionary history of the Australasian and Pacific region are generally supported by genetic analyses. There is substantial evidence that aboriginal Australians and the highlanders of New Guinea are more closely related to each other than either is to other Pacific and Asian populations

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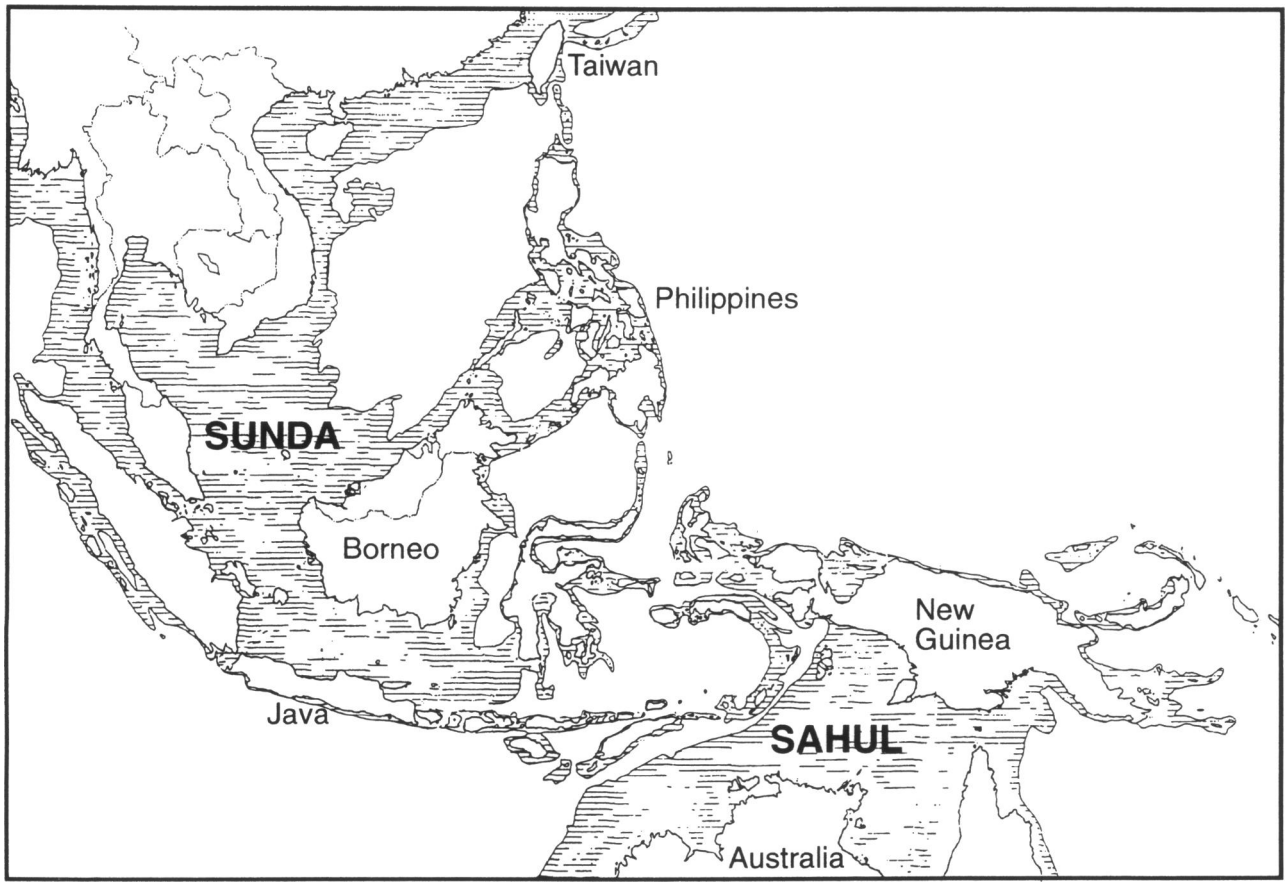


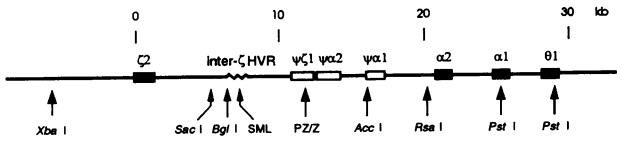
Figure 1 Southeast Asia and northern Australia, showing the boundaries of the ancient landmasses of Sunda and Sahul. The shaded areas indicate the extent of the land exposed during the Pleistocene.

(Hill and Serjeantson 1989; Nei and Roychoudhury 1993) and that they have been isolated both from Asia and from each other for a long time. In world surveys of protein loci, both these populations have the lowest heterozygosities and a number of novel mutations (Kirk 1989).

Nonetheless, the relationship between the aboriginal populations of New Guinea, Australia, and island Southeast Asia is not entirely clear. In a study of protein variation, Kirk (1989) reported a closer affinity between aboriginal Australians and Southeast Asians than between aboriginal Australians and New Guineans. A lack of association between aboriginal Australians and New Guinea highlanders was also observed for mtDNA (Stoneking and Wilson 1989; Stoneking et al. 1990) and in early studies on allelic variation at the HLA system (Serjeantson 1985). More recent studies of HLA-DR haplotypes, however, do indicate close associations between aboriginal Australians and New Guineans (Gao and Serjeantson 1991), a relationship that is also supported by analyses of other nuclear DNA RFLPs (Chen et al. 1990*b*, 1992; Lin et al. 1994).

Data obtained from the globin gene complexes have so far been equivocal. A study of β-globin RFLP haplotypes in the region (Chen et al. 1990*a*) was uninformative of the pattern of affinities among these populations. A study of α-globin haplotypes of Australians from the northwest coast (Tsintsof et al. 1990) argued against a substantial common ancestral link with New Guinea highlanders and concluded that there had been multiple colonizing events of Australia. However, in this study, the possibility of recent Southeast Asian admixture was not excluded.

In the present study, we have characterized the α-globin gene complex of Australians from one tribe in central Australia. In contrast to Tsintsof et al. (1990), we find convincing evidence of strong links with New Guinea highlanders, implying that migrations into ancient Sahul originated from a single source population. We suggest that more recent gene flow from island Southeast Asia, resulting in the aboriginal populations on of the north and northwest coasts of Australia becoming admixed, explains some of the ambiguous results of previous studies.



Ia	+	+	-	M	PZ	+	+	-	-
IIe	+	-	-	S	PZ	+	-	-	-
IIIa	-	-	+	M	Z	-	-	-	-
IIIb	+	-	+	M	Z	-	-	-	-
IVa	+	-	-	S	PZ	+	-	+	+
IVb	-	-	-	S	PZ	+	-	+	+
IIh	-	-	+[1]	M	PZ	+	-	-	-
IIi	-	+	+	M	PZ	+	-	-	-
IIj	-	+	-	M[2]	PZ	+	-	-	-
VIc	-	-	+	M	PZ	-	-	-	-
VId[3]	+	-	-	S	PZ	-	-	-	-
VIIb	-	-	+	M	Z	+	-	-	-
IXa	-	-	+	M	Z	+	+	-	-

Figure 2 α-Globin complex, showing the functional genes (filled boxes) and pseudogenes (empty boxes), along with the approximate position of polymorphic sites and the restriction enzymes used to reveal the site. Six haplotypes—Ia, IIe, IIIa, IIIb, IVa, and IVb—present in the central Australian population have been described by Higgs et al. (1986). The seven new haplotypes observed in this study are shown below the line. (1) The IIh *BglI* fragment was slightly larger than that present on other *BglI*+ haplotypes; (2) some examples of this haplotype had a slightly smaller interzeta HVR "M" allele than normal; and (3) this haplotype carries a novel *AccI* site polymorphism (see text).

Methods

Blood samples were collected with informed consent from 119 members of the Wailbri tribe at Yuendumu located on the edge of the Tanami Desert, ~300 km northwest of Alice Springs in central Australia. Records from the tribe have been kept for the past 30 years, so the family histories of most individuals were known.

The blood was collected into heparin and frozen at −20°C. DNA was prepared by Proteinase-K digestion followed by phenol-chloroform extraction, using an Applied Biosystems Model 340A nucleic acid extractor, and diluted to 1 μg/3 μl with 10 mM Tris-Cl, 1 mM EDTA (pH 7.4).

Genotyping of $-\alpha$ and $\alpha\alpha\alpha$ rearrangements and subtyping of $-\alpha^{3.7}$ deletions was determined as described by O'Shaughnessy et al. (1990). It was possible to determine the α-globin genotype for each of the 119 samples collected, but only 108 samples provided sufficient material for complete haplotype analysis.

Determination of the α-globin haplotypes was carried out as described by Higgs et al. (1986) (fig. 2). The haplotypes present in an individual can be resolved if all RFLP sites are homozygous or if only one site is heterozygous. If an individual is heterozygous at more sites, the haplotype can only be assigned with confidence by using additional information, such as genealogical data, that allows the coinheritance of linked RFLPs to be followed (Martinson et al. 1995). In samples where one or a small number of haplotypes makes up the majority of the observed repertoire, it is often possible to resolve the haplotypes of some multiply heterozygous individuals by subtraction of the common haplotype (Tsintsof et al. 1990; Martinson et al. 1994), a technique that has also been used in the determination of HLA haplotypes for populations in this region (Gao and Serjeantson 1991). Over 50% of the individuals studied here were homozygous for the IIIa haplotype, with its distinguishing "Z" allele. Compound heterozygotes containing the Z polymorphism could thus usually be resolved into constituent haplotypes, which makes the subtraction approach very effective for this particular sample set. Only 2% of the samples remained unclassified.

Results

The α-globin haplotypes of the 106 central Australians (of a total of 108 sampled) that were successfully characterized are given in table 1 and figure 2. Of the 212 chromosomes, 158 (74.5%) were group III haplotypes, of which 157 were type IIIa (−−+MZ−−−−) and one chromosome was type IIIb. This high frequency of the IIIa haplotype was instrumental in the successful resolution of a large proportion (98%) of the haplotypes in the total sample (see Methods).

A total of seven different previously unreported hap-

Table 1

α-Globin Haplotypes in Central Australians

Haplotype	No.
Ia	1
IId	1
IIe	16
IIh[a]	5
IIi[a]	1
IIj[a]	157
IIIb	1
IVa	10
IVb	2
VIc[a]	1
VId[a]	7
VIIb[a]	3
IXa[a]	2
Total	212

[a] Haplotypes not previously described.
[b] Including seven rearrangements.

lotypes were found in this Australian population (fig. 2), of which six (IIh, IIi, IIj, VIc, VId, and VIIb) are new subtypes (as defined by the four 5′ sites) of existing groups, and one (−−+MZ++−−) is characterized by a new 3′ motif, Z++−− (group IX; fig. 2).The seven chromosomes with the VId haplotype had a novel *Acc*I polymorphism, which resulted in a fragment that was only differentiated from the *Acc*I (+) fragment characteristic of typical group II haplotypes because its size was slightly larger. However, analysis of the "normal" *Acc*I site by PCR (Zago et al. 1995) clearly showed that it was absent, specifying these haplotypes as group VI. Thus, although, *Acc*I (−), these chromosomes can easily be mistaken for group II (*Acc*I+) haplotypes if assayed by low-resolution Southern blotting.

The new haplotypes IIh, VIc, and VIIb, share the −−+M 5′ motif that is characteristic of the predominant IIIa haplotype, and the VId haplotype has the same 5′ sites as the second-most-common haplotype, IIe, suggesting these new variants (with the exception of the VId) have been generated by recombination in the interzeta hypervariable region or pseudozeta/zeta gene intron regions between the IIIa or IIe and other haplotypes.

In the central Australian sample, eight chromosomes, from 238 typed, had α-gene rearrangements: seven chromosomes (3.3%) had the $-\alpha^{3.7}$I deletion, and one (0.5%) had the ααα rearrangement. All of the $-\alpha^{3.7}$I deletions were IIIa haplotypes. This compares with the study by Tsintsof et al. (1990), which reported a 3.4% gene frequency of $-\alpha^{3.7}$, of which 90% were subtype I and 10% were $-\alpha^{3.7}$III, 0.7% ααα, and 2.4% of the triplicated ζ gene (ζζζ) arrangement, and that of Yenchitsomanus et al. (1986*a*), who found the $-\alpha^{3.7}$ deletion (subtypes I and II) occurring at frequencies of 1.4%, 3.9%, and 13.4% in three areas on the north and northwest coast of Australia, but with no deletions observed in the sample of 22 individuals from central Australia. The blood samples used for that study had been collected on various occasions over a period of 15 years, and there does not appear to be any record of the individuals sampled. In New Guinea highlanders, Flint et al. (1986) reported that the incidence of $-\alpha^{3.7}$ was 0.8%, with 3% $-\alpha^{4.2}$ and 1% ααα. The gene deletions were associated with type IIIa haplotypes or a IIIa variant. Yenchitsomanus et al. (1986*b*), found 8% $-\alpha^{3.7}$, all subtype I, 3% $-\alpha^{4.2}$, and 0.4% ααα.

The composition of normal α-globin haplotypes for the central Australian sample is compared with previously published data for northwestern Australians and other Melanesian, Southeast Asian and Oceanic populations (table 2). It is clear that there is generally a good correspondence between the central Australian and the New Guinea data, with comparable high frequencies of group III haplotypes. These results are in contrast to the aboriginal Australian data of Tsintsof et al. (1990), in which there were substantial proportions of group I and group II haplotypes, in addition to group III.

Chord distances (Cavalli-Sforza and Edwards 1967) between all pairs of populations were computed using NTSYS-pc (Rohlf 1993), and a neighbor-joining tree (fig. 3) was constructed using MEGA (Kumar et al. 1993). Figure 3 shows a close relationship between aboriginal central Australians and Melanesians (fig. 3). There is a major split dividing the Australasian and island Melanesian populations from the Polynesians (Samoa and Tonga) and Taiwanese. The position of the northwestern Australians in this tree reflects admixture from a Southeast Asian source.

Patterns of population structure were investigated by using an analysis of pairwise population relationships that was suggested by Hudson et al. (1992). Table 3 presents the genetic differentiation statistics for pairs of populations, computed from haplotypes (H_{ST}) and haplotypes as sequences (K_{ST}), and the probabilities of observing the average within-population diversity statistics, H_S and K_S, from 2,000 random partitions of the combined populations. $H_{ST} = 1 - (H_S/H_T)$ and $K_{ST} = 1 - (K_S/K_T)$, where K_T and H_T are estimates of total diversity in the combined population. When >5% of random partitions gave diversities within populations as small as, or smaller than, the observed partition, the combined population was considered not to be differentiated. The central Australian sample is significantly differentiated from all others, but least so from the highlands of Papua New Guinea. Two pairs of populations do not show significant differentiation, and these results are probably due to the small sample sizes available for distinguishing Tonga from Taiwan and Vanuatu from Fiji.

The haplotype frequency distributions observed within each sample were further examined for concordance with the frequency distributions expected given counts of haplotypes observed, when a history of unique mutation and recombination events, random mating, and constant population size are assumed (Ewens 1972; Watterson 1978). Only the haplotype frequency distribution for the central Australian sample was found to depart significantly from expected (Clark 1987). The observed and expected homozygotes and numbers of haplotypes for all populations are given in table 4. The distinctive frequency distribution of the central Australian sample is characterized by the very high frequency of type IIIa and wide range of low-frequency haplotypes, which are not found in the other populations. We suggest that this distribution reflects recovery from a bottleneck in the founding migration to Australasia. The higher heterozygosity and equilibrium frequency distribution of Australians from the northwest coast is probably due to gene flow from Southeast Asia.

Table 2

α-Globin Haplotypes of Central Australians Compared to Populations from Northwest Australia, Melanesia, Southeast Asia, and Oceania

Haplotype	Subtype	Group	Central Australia[a]	Northwest Australia[b]	New Guinea Highland[c]	New Guinea Coast[c]	Vanuatu[c]	Fiji[c]	Tonga[d]	Samoa[d]	Taiwan[e]	
Ia	++−M	PZ++−−	1	20	...	1	11	13	11	80	29	Ia
Ib	−+−M	PZ++−−	...	3	...	...	...	...	...	2	4	Ib
Ia/b	+/−+−M	PZ++−−	...	3	...	...	...	...	...	...	...	Ia/b
Ic	+−−M	PZ++−−	...	...	...	...	...	1	...	...	...	Ic
Id	−+−L	PZ++−−	...	...	...	...	...	1	...	...	3	Id
IIa	−+−L	PZ+−−−	...	16	...	1	...	...	5	13	14	IIa
IIb	++−L	PZ+−−−	...	2	...	...	...	...	...	...	1	IIb
IIc	++−M	PZ+−−−	...	1	...	5	...	...	1	9	6	IIc
IId	−−−S	PZ+−−−	1	...	...	1	3	7	...	46	3	IId
IIe	+−−S	PZ+−−−	16	6	1	12	...	...	4	38	13	IIe
IIf	+−+S	PZ+−−−	...	...	...	...	...	2	...	...	2	IIf
IIg	+−−M	PZ+−−−	...	...	...	...	...	...	...	...	3	IIg
IIh	−−+M	PZ+−−−	5	...	...	3	...	...	...	...	...	IIh
IIi	−++M	PZ+−−−	1	...	...	1	...	...	...	...	...	IIi
IIj	−+−M	PZ+−−−	5	...	...	1	...	...	...	...	...	IIj
IIIa	−−+M	Z−−−−	157	61	77	41	13	13	8	60	4	IIIa
IIIb	+−+M	Z−−−−	1	8	6	1	4	...	...	...	...	IIIb
IIIa/b	+/−−+M	Z−−−−	...	3	...	...	...	...	...	...	...	IIIa/b
IIIc	−+−L	Z−−−−	...	...	1	...	...	...	...	...	...	IIIc
IIIe	−+−M	Z−−−−	...	...	...	...	...	2	...	5	...	IIIe
IIIf	−−−M	Z−−−−	...	...	2	...	...	...	...	...	...	IIIf
IIIh	+++M	Z−−−−	...	...	...	1	...	...	...	5	2	IIIh
IVa	+−−S	PZ+−++	10	11	49	29	16	10	...	46	...	IVa
IVb	−−−S	PZ+−++	2	1	3	...	1	...	...	...	...	IVb
IVc	−+−L	PZ+−++	...	2	...	...	...	...	...	...	...	IVc
Vb	−−+M	PZ+−−+	...	...	...	2	...	2	...	1	...	Vb
Vc	+−−S	PZ+−−+	...	...	1	43	...	8	...	1	...	Vc
Vd	−−−S	PZ+−−+	...	...	...	6	...	...	...	...	...	Vd
VIc	−−+M	PZ−−−−	1	...	...	...	...	...	...	...	...	VIc
VId	+−−S	PZ−−−−	7	...	1	...	...	...	...	...	...	VId
VIIb	−−+M	Z+−−−	3	...	...	2	...	...	...	...	...	VIIb
VIIIa	+−−S	PZ+−+−	...	...	1	...	...	...	...	...	...	VIIIa
IXa	−−+M	Z++−−	2	...	...	...	...	...	...	...	...	IXa
Xa	−−+M	Z−+−−	...	2	...	...	...	...	...	...	...	Xa
Sum classified			212	139	142	150	48	59	29	306	84	
Unclassified			4	23	38	52	0	19	35	4	80	
Total			216	162	180	202	48	78	64	310	164	

[a] Present study.
[b] Tsintsof et al. (1990).
[c] O'Shaughnessy et al. (1990), Martinson et al. (1994), and authors' unpublished data.
[d] Hertzberg et al. (1988) and O'Shaughnessy et al. (1990).
[e] Authors' unpublished data.

Discussion

This study demonstrates that the α-globin haplotype composition of central Australians is similar to those of the coastal and highlands populations of New Guinea. All these populations feature high frequencies of the IIIa haplotype and the presence of IVa and IVb haplotypes. The distribution of group III and IV haplotypes is relatively restricted outside of Australia and Melanesia, although they are found in some African populations. The central Australian population is closest to the New Guinea highlanders, consistent with other genetic and archaeological data suggesting that the early occupation of Sahul included both Australia and New Guinea and that these people migrated from a common Southeast Asian source.

That ancestral source population must have been quite distinct in α-haplotype composition from the contemporary populations of island and mainland Southeast Asia, which are distinguished by a predominance of group I and II haplotypes (and relatively few (<10%) of groups III and IV) (O'Shaughnessy et al. 1990). The ancestors of present-day Austronesian-speaking inhabitants of the Indo-Malaysian archipelago probably arrived via the Philippines from the north within the past 5,000 years (Bellwood 1985). Our analysis thus precludes any major colonizing events into Australia after that date.

That there has been contact between the Indo-Malaysian archipelago and Australia is not disputed. The α-haplotype composition of aboriginal Australians from the northwest coast is clearly different from the central Aus-

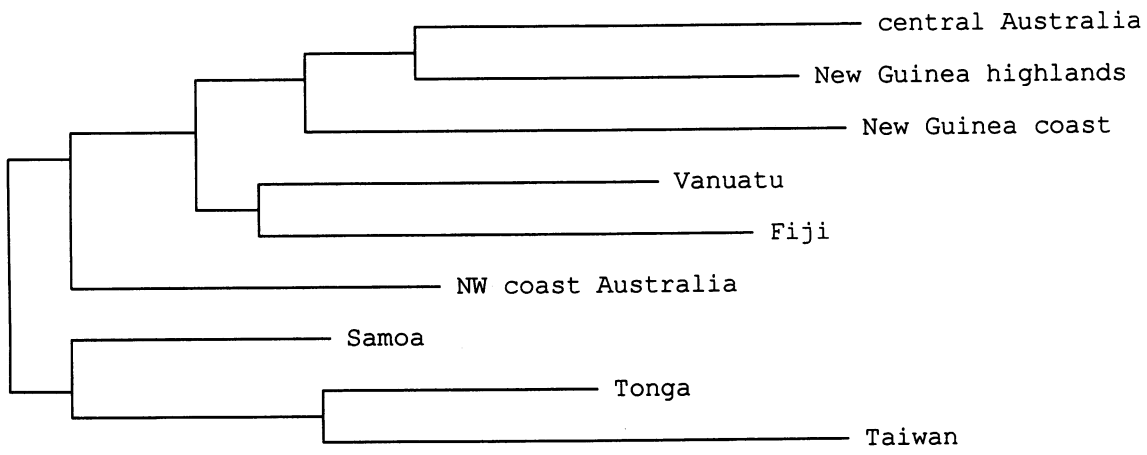


Figure 3 Neighbor-joining tree (Saitou and Nei 1987) for nine populations, based on chord distances (Cavalli-Sforza and Edwards 1967)

tralian population, with a substantial proportion of Southeast Asian group I and II haplotypes (table 2). Archaeological studies have shown that, during the past few centuries at least, the north and northwest coast of Australia has been visited by fishermen and others from what is now Indonesia (Berndt and Berndt 1985, pp. 1–24). Gene flow is evident from previous genetic analyses, especially for individuals from coastal regions, and probably accounts for the much wider variety of haplotypes and globin gene rearrangements found by Tsintsof et al. (1990) and Yenchitsomanous et al. (1986*a*). The central Australians, by contrast, have negligible levels (1/200) of the most common haplotypes that characterize Southeast Asians, and, conversely, the rare haplotype groups found in central Australians have never been observed in Southeast Asia (authors' unpublished observations). All these findings imply that central Australia has remained isolated from island Southeast Asia for many millenia.

Although the incidence of α-thalassaemia in Australians is low (~3% in two studies) and most probably reflects the lack of malaria in their recent historic environment, this frequency of α-gene deletions is still higher than in nonmalarial regions such as Britain and Iceland, where such deletions are virtually absent. It is likely that there has been malaria on the northern coast of Australia (Groube 1993), and migrations into central regions from the coast could have introduced α-thalassaemia into the interior. The $-\alpha^{3.7}$I deletions in central Australians clearly have a different origin from the majority of New Guinea deletions found in both north coastal and highland samples (which are either $-\alpha^{3.7}$ subtype III or $-\alpha^{4.2}$ [Flint et al. 1986]). Yenchitsomanus et al. (1986*b*) did however report $-\alpha^{3.7}$I in the isolated Chimbu region of the New Guinea highlands, a deletion that probably has arisen locally, and has not been brought in from Southeast Asia in more recent years. Moreover, on the north coast of Australia, $-\alpha^{3.7}$I deletions are associated with haplotypes Ia, IIa, and IIIa. It seems likely that the former two are Southeast Asian variants (and thus imports), while the latter could be Australian or New Guinean. New Guinea also seems the more probable source of the rare examples of $-\alpha^{3.7}$III (which probably originated in northern Melanesia [Flint et al. 1986]) encountered in northern Australia.

Table 3

Analysis of Population Differentiation

	H_{ST}	Probability (H_S)	K_{ST}	Probability (K_S)
Central Australia + New Guinea highland	.06	.000	.07	.000
Central Australia + Coastal New Guinea	.12	.000	.21	.000
Central Australia + Vanuatu	.10	.000	.12	.000
Central Australia + Fiji	.10	.000	.16	.000
Central Australia + NW Australia	.05	.000	.06	.000
Central Australia + Samoa	.13	.000	.19	.000
Central Australia + Tonga	.08	.000	.12	.000
Central Australia + Taiwan	.19	.000	.33	.000
New Guinea highland + Coastal New Guinea	.06	.000	.07	.000
Vanuatu + Fiji	.01	.077 (ns)	<.01	.401 (ns)
Tonga + Taiwan	.01	.132 (ns)	.01	.057 (ns)
Samoa + Taiwan	.02	.000	.03	.000

Table 4

Test of Goodness-of-Fit to Equilibrium Distributions of Haplotypes

		No. of Haplotypes		Homozygosity		
	No.	Observed	Expected	Observed	Expected	Probability
Central Australia	212	14	5.0	.56	.23	.004
Northwest Australia	133	12	11.7	.26	.24	.328
New Guinea, highland	142	10	7.1	.41	.29	.129
New Guinea, coast	150	16	15.2	.20	.18	.287
Vanuatu	48	6	9.6	.23	.37	.900
Fiji	59	10	14.4	.15	.22	.886
Tonga	29	5	7.7	.24	.37	.897
Samoa	306	12	21.0	.17	.28	.934
Taiwan	84	12	14.3	.18	.21	.576

Both aboriginal Australians and New Guinea highlanders have notably low genetic diversity at α-globin and other loci, and this has been interpreted in previous studies as a result of isolation and long-term small population sizes. However, it is possible that the original diversity was lost in a population bottleneck prior to or during the occupation of Sahul. Also, it is likely that the genetic differences found between central Australians and the New Guinea highlanders are due to the accumulation of new haplotypes, generated by mutation and recombination, rather than to drift in generational sampling, as would be expected if population sizes remained small. There is no evidence that these rare haplotypes derive from populations now settled in island Southeast Asia. The presence of locally specific haplotypes suggests moderate population sizes and low rates of gene flow among these regions. The nonequilibrium distribution for the central Australian aboriginal sample is consistent with population expansion, generated by natural recruitment rather than immigration. Following the bottleneck that occurred in an ancestral population of both Australia and New Guinea, diversity has been recovering by the incorporation of new haplotypes.

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