

haplogroup M was brought to Asia from East Africa, along the southern route, by the earliest migration wave of anatomically modern humans, ~60,000 years ago (Kivisild et al. 1999a, 1999b, 2000; Quintana-Murci et al. 1999). Another deep late Pleistocene link through haplogroup U was found to connect western Eurasian and Indian populations. Less than 10% of the maternal lineages of the caste populations had an ancestor outside India in the past 12,000 years (Kivisild et al. 1999a, 1999b). mtDNA profiles from a larger set of populations all over the subcontinent have bolstered the view of fundamental genomic unity of Indians (Roychoudhury et al. 2001). In contrast, the Y-chromosome genetic distance estimates showed that the chromosomes of Indian caste populations were more closely related to Europeans than to eastern Asians (Bamshad et al. 2001). The tendency of higher caste status to associate with increasing affinities to European (specifically to eastern European) populations hinted at a recent male-mediated introduction of western Eurasian genes into the Indian castes' gene pool. The similarities with Europeans were specifically expressed in substantial frequencies of clades J and R1a (according to Y Chromosome Consortium [YCC] 2002 nomenclature) in India. The exact location of the origin of these haplogroups is still uncertain, as is the timing of their spread (Zerjal et al. 1999; Bamshad et al. 2001; Passarino et al. 2001; Quintana-Murci et al. 2001; Wells et al. 2001).

To address the question of the origin of Indian maternal and paternal lineages further, we analyzed variation in mtDNA, the Y chromosome, and one autosomal locus (Jin et al. 1999) in two southern Indian tribal groups from Andhra Pradesh and compared them with Indian caste groups and populations from Iran, the Middle East, Europe, and central Asia.

Material and Methods

Chenchus were first described as shy hunter-gatherers by the Mohammedan army in 1694. They reside in the ranges of Amrabad Plateau, Andhra Pradesh, and have a population size of ~17,000. Their society is patriarchal and patrilineal, with marriage occurring mostly between clans (kulam) of equal status. Chenchus are described as an australoid population, when physical anthropological features are used as criteria (Bhowmick 1992; Singh 1997; Thurin 1999). The Chenchu language belongs to the Dravidian language family.

More than 300,000 Koyas live in the plains and forests on both sides of the Godavari River in Andhra Pradesh. Their language is related to the Gondi, which connects a large group of Dravidian languages in southern India. They are primarily farmers and live in villages. Exoga-

mous patrilineal clans make up their social structure, as they do for the Chenchus (Singh 1997).

After informed consent was obtained, 180 blood samples were collected from healthy and maternally unrelated volunteers belonging to Chenchu and Koya tribes from Andhra Pradesh, 106 West Bengalis of different caste ranks, 58 Konkanastha Brahmins from Bombay, and 53 Gujaratis; in addition, 132 samples were collected from Sri Lanka (including 40 Sinhalese), 112 from Punjabis of different caste rank, and 139 samples from Uttar Pradesh (including those described by Kivisild et al. 1999a). The Lambadi ($n = 86$) sample from Andhra Pradesh, the Boksas ($n = 18$) from Uttar Pradesh, and the Lobanas ($n = 62$) from Punjab are described by Kivisild et al. (1999a) and were further analyzed here for Y-chromosomal and additional mtDNA markers. The Y-chromosome sample size of each population is shown in figure 3. In addition, 388 Turks from central Turkey (Cappadocia), 202 Kuwaitis, 202 Saudi Arabians, and 440 Iranians were used in mtDNA haplogroup frequency comparisons. In addition, Y-chromosome STR data from six loci were used for comparing intra- and interhaplogroup variances in selected haplogroups and populations. These included 88 central Asians (Altai, Kirghiz, Uzbek, and Tajik) belonging to haplogroup R1a, and Y chromosomes from haplogroups I (12 Estonians and 9 Czechs), J (6 Czechs), and R1 (39 Estonians and 30 Czechs). Further details about these populations will be published elsewhere. DNA was extracted using standard phenol-chloroform methods (Sambrook et al. 1989).

D A A —

Hypervariable segments (HVS) I (nucleotide positions [nps] 16024–16400) and II (nps 16520–300) of the control region were sequenced in 96 Chenchu and 81 Koya samples. In addition, three segments of the coding region (nps 1674–1880, 4761–5260, and 8250–8710) were sequenced, and informative RFLP positions (Macaulay et al. 1999; Quintana-Murci et al. 1999) were checked (table 1) in selected individuals from different haplotypes, to define haplogroup affiliations. Published HVS-I sequence data used for haplotype comparisons included 250 Telugus from Andhra Pradesh (Kivisild et al. 1999a; Bamshad et al. 2001), 48 Haviks, 43 Mukris, and 7 Kaders from Kerala and Karnataka (Mountain et al. 1995).

-C — A —

Y-chromosomal haplogroups were determined by RFLP and denaturing high-performance liquid chromatography (dHPLC) methods, using 35 biallelic markers (Rosser et al. 2000; Underhill et al. 2000, 2001b) that are shown in hierarchical relation to one another

in figure 3. Length variation at six STR loci (DYS19, DYS388, DYS390, DYS391, DYS392, and DYS393) was typed using Cy5-labeled primers, and amplification products were subjected to electrophoresis on ALF Express (Pharmacia-Amersham). Scoring of repeat lengths was standardized by use of controls sequenced by P. de Knijff.

1.

A 246-bp segment of the MX1 locus of chromosome 21, containing eight polymorphic sites in humans, was sequenced, and a nearby *StuI* recognition site polymorphism was determined according to Jin et al. (1999) in 42 Chenchus, 28 Koyas, 35 West Bengalis, 34 Punjabis, and 35 Turks from Cappadocia.

2.

Preparation of sequencing templates was performed according to methods described by Kaessmann et al. (1999). Purified products were sequenced with the DYE-namic ET terminator cycle sequencing kit (Amersham Pharmacia Biotech) and were analyzed on an ABI 377 DNA Sequencer. Sequences were aligned and analyzed with the Wisconsin Package (GCG).

D. A.

Median networks (Bandelt et al. 1995, 1999) were constructed using the Network 2.0 program (A. Röhl; Shareware Phylogenetic Network Software Web site) with default settings. Cluster ages were calculated using ρ , the averaged distance to a specified founder haplotype, according to Forster et al. (1996), as well as standard errors as described by Saillard et al. (2000a). In mtDNA coalescent calculations, using the estimator ρ , we use a mutation rate of one transition in the segment between nps 16090 and 16365 per 20,180 years, calibrated with the inference that links Eskimo-Aleutian haplogroup A diversity to their post-Younger Dryas population expansion (Forster et al. 1996).

To calculate the 95% credible regions (CR85(W)r/F2-345-ucaoCR85(W,)]T485(trl20(wA30.grages)-u-475(po1aration)-41 Tc[(,)

Table 1

mtDNA Haplotypes in Chenchu and Koya Populations

[illegible]

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Table 2**Major mtDNA Lineage Clusters in India and Western Asia**

		NO. OF LINEAGES IN CLUSTER (95% CR FOR PROPORTION) ^a										
POPULATION (n)	D ^b	L1–L3	M	M2	M3	M5	MΔ9bp	U2i, U7	U1, U3–U6, U*	HV ^c , TJ, N1, X	B, F ^d	R*
Tribal:												
Chenchus (96) ^e	.87	0 (.00–.03)	93 (.91–.99)	17 (.11–.27)	1 (.00–.06)	18 (.12–.28)	3 (.01–.09)	0 (.00–.03)	0 (.00–.03)	0 (.00–.03)	0 (.00–.03)	1 (.00–.06)
Koyas (81) ^e	.94	0 (.00–.04)	56 (.58–.78)	15 (.12–.28)	5 (.03–.14)	0 (.00–.04)	17 (.14–.31)	1 (.00–.07)	0 (.00–.04)	0 (.00–.04)	0 (.00–.04)	25 (.22–.42)
Tamil Nadu (49) ^f	.96	0 (.00–.06)	35 (.58–.82)	1 (.01–.11)	12 (.15–.38)	0 (.00–.06)	0 (.00–.06)	6 (.06–.24)	2 (.01–.14)	0 (.00–.06)	0 (.00–.06)	6 (.06–.24)
Western Bengal (34) ^f	.99	0 (.00–.08)	22 (.48–.79)	2 (.02–.19)	3 (.03–.23)	0 (.00–.08)	0 (.00–.08)	7 (.10–.37)	0 (.00–.08)	0 (.00–.08)	0 (.00–.08)	5 (.07–.30)
Caste:												
Western Bengalis (106) ^e	.97	0 (.00–.03)	76 (.63–.79)	4 (.02–.09)	7 (.03–.13)	6 (.03–.12)	0 (.00–.03)	10 (.05–.17)	1 (.01–.05)	6 (.03–.12)	0 (.00–.03)	12 (.07–.19)
Gujaratis and Konkanastha Br. (111) ^e	.99	0 (.00–.03)	53 (.39–.57)	5 (.02–.10)	7 (.03–.13)	0 (.00–.03)	0 (.00–.03)	20 (.12–.26)	5 (.02–.10)	11 (.06–.17)	5 (.02–.10)	12 (.06–.18)
Kerala/Karnataka (99) ^g	.96	0 (.00–.03)	63 (.54–.72)	15 (.09–.24)	6 (.03–.13)	15 (.09–.24)	0 (.00–.03)	20 (.14–.29)	1 (.01–.05)	...	0 (.00–.03)	9 (.05–.16)
Lambadis (86) ^h	.99	0 (.00–.03)	55 (.53–.73)	9 (.06–.19)	4 (.02–.11)	9 (.06–.19)	0 (.00–.03)	9 (.06–.19)	2 (.01–.08)	7 (.04–.16)	0 (.00–.03)	11 (.07–.22)
Lobanas (62) ^h	.98	0 (.00–.05)	34 (.43–.67)	3 (.02–.13)	3 (.02–.13)	5 (.04–.18)	0 (.00–.05)	2 (.01–.11)	1 (.00–.09)	5 (.04–.18)	0 (.00–.05)	11 (.10–.29)
Punjabis (112) ^e	.99	0 (.00–.03)	46 (.32–.50)	1 (.00–.05)	5 (.02–.10)	1 (.00–.05)	0 (.00–.03)	15 (.08–.21)	8 (.04–.14)	21 (.13–.27)	6 (.03–.11)	11 (.06–.17)
Sri Lanka (132) ^e	.99	0 (.00–.02)	77 (.50–.66)	9 (.04–.13)	6 (.02–.10)	2 (.01–.05)	0 (.00–.02)	19 (.09–.21)	5 (.02–.09)	11 (.05–.14)	2 (.01–.05)	18 (.09–.21)
Telugu, upper (59) ⁱ	.99	0 (.00–.05)	36 (.48–.72)	3 (.02–.14)	11 (.11–.30)	0 (.00–.05)	0 (.00–.05)	10 (.10–.29)	1 (.00–.09)	2 (.01–.11)	0 (.00–.05)	9 (.08–.27)
Telugu, middle (114) ^j	.99	0 (.00–.03)	73 (.55–.72)	7 (.03–.12)	4 (.01–.09)	5 (.02–.10)	0 (.00–.03)	10 (.05–.15)	1 (.01–.05)	6 (.03–.11)	0 (.00–.03)	24 (.15–.29)
Telugu, lower (70) ^j	.99	0 (.00–.04)	50 (.60–.81)	7 (.05–.19)	1 (.00–.08)	3 (.02–.12)	0 (.00–.04)	5 (.03–.16)	0 (.00–.04)	1 (.00–.08)	0 (.00–.04)	15 (.14–.32)
Uttar Pradesh (139) ^{e,h}	.99	0 (.00–.02)	79 (.49–.65)	4 (.01–.07)	14 (.06–.16)	0 (.00–.02)	0 (.00–.02)	21 (.10–.22)	3 (.01–.06)	9 (.04–.12)	2 (.00–.05)	20 (.10–.21)
West Asian:												
Iranians (440) ^e	.99	2 (.00–.02)	24 (.04–.08)	0 (.00–.01)	5 (.01–.03)	0 (.00–.01)	0 (.00–.02)	41 ^j (.07–.12)	90 (.17–.25)	245 (.51–.60)	2 (.00–.02)	17 (.02–.06)
Turks, Cappadocia (388) ^e	.99	1 (.00–.01)	16 (.03–.07)	0 (.00–.01)	0 (.00–.01)	0 (.00–.01)	0 (.00–.01)	7 (.01–.04)	93 (.20–.29)	244 (.58–.68)	1 (.00–.01)	7 (.01–.04)
Middle East (406) ^{e,k}	.99	41 (.08–.13)	30 (.05–.10)	2 (.00–.02)	1 (.00–.01)	0 (.00–.01)	0 (.00–.03)	15 (.02–.06)	40 (.07–.13)	269 (.62–.71)	1 (.00–.01)	3 (.00–.02)

^a U2i excludes variants of U2 with 16129C; U* = other derivatives of haplogroup U; R* = derivatives of haplogroup R that do not belong to HV, TJ, U, B, and F.

^b HVS-I haplotype diversity.

^c Including pre-HV, as defined by Saillard et al. 2000b.

^d None of the Indian samples belonged to haplogroup B.

^e From the present study.

^f From Roychoudhury et al. 2001.

^g From Moutain et al. 1995.

^h From Kivisild et al. 1999.

ⁱ From Bamshad et al. 2001.

^j Forty Iranians belonged to U7 and one belonged to U2i.

^k The Middle East sample includes 202 Kuwaitis and 204 Saudi Arabians.



Figure 2 A network of haplogroup M2 haplotypes. Circle areas are proportional to haplotypes frequencies. Variant bases are numbered

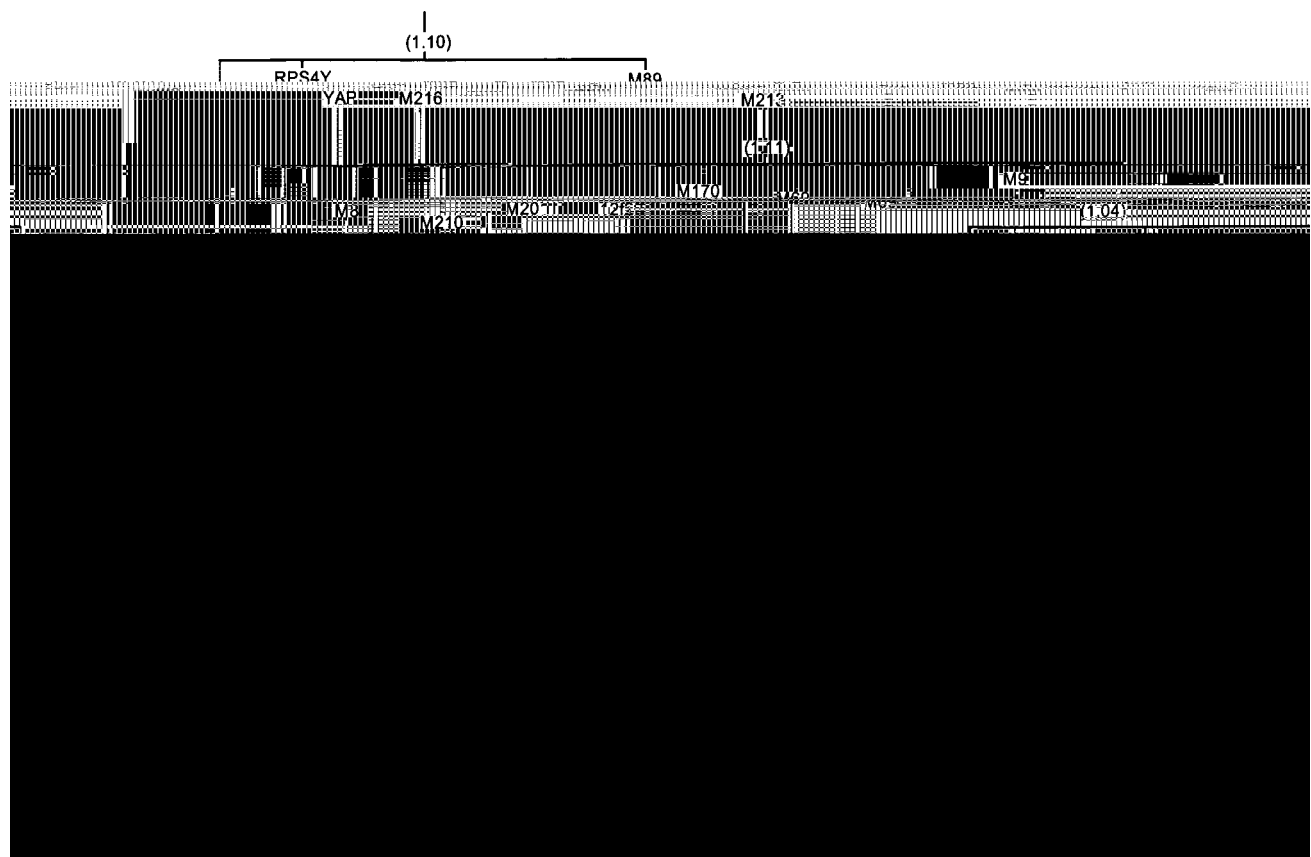


Figure 3 Y-chromosomal SNP tree and haplogroup frequencies in 8 Indian populations. Haplogroup defining markers (and their background average variances of 6 STR loci) are shown along the branches of the tree.

Table 3**Major Y-Chromosomal Haplogroups in India Compared with Western Eurasia**

	FREQUENCY (95% CR FOR PROPORTION)			
POPULATION (n)	H1 (M52)	L (M11)	R2 (M124)	REFERENCE
India:				
Punjab (66)	.03 (.01–.10)	.12 (.06–.22)	.05 (.02–.13)	Present study
West Bengal (31)	.10 (.04–.25)	.00 (.00–.09)	.23 (.12–.40)	Present study
AP tribes (82)	.49 (.43–.64)	.07 (.04–.15)	.04 (.01–.10)	Present study
AP tribes (67)	.10 (.05–.20)	.04 (.02–.12)	...	Ramana et al. 2001
AP castes (125)	.10 (.06–.16)	.08 (.04–.14)	...	Ramana et al. 2001
Tamil Nadu (259)	.17 (.13–.22)	.29 (.24–.35)	.07 (.05–.11)	Wells et al. 2001
Tadjikistan (168)	.01 (.00–.04)	.11 (.07–.16)	.06 (.03–.11)	Wells et al. 2001
Uzbeks (366)	.02 (.01–.04)	.03 (.02–.05)	.02 (.01–.04)	Wells et al. 2001
Kyrgyzstan (92)	.00 (.00–.03)	.00 (.00–.03)	.02 (.01–.08)	Wells et al. 2001
Kazakstan (95)	.01 (.00–.06)	.01 (.00–.06)	.01 (.00–.06)	Wells et al. 2001
Iran (52)	.00 (.00–.06)	.04 (.01–.13)	.02 (.01–.10)	Wells et al. 2001
Near East (101)	.01 (.00–.05)	.02 (.01–.07)	.00 (.00–.03)	Semino et al. 2000
Caucasus (147)	.00 (.00–.02)	.01 (.00–.05)	.00 (.00–.02)	Wells et al. 2001
Europe (839)	.00 (.00–.01)	.01 (.00–.01)	.00 (.00–.01)	Semino et al. 2000

(table 2). Furthermore, 32% of the Koya M* HVS-I sequences shared an A at hypervariable np 16129, which is characteristic of a likely polyphyletic HVS-I clade M5 (Bamshad et al. 2001). The loss of 12403 *MnII*, one of the four defining markers of African M1 cluster (Maca-Meyer et al. 2001), was not found in either tribal sample (table 1).

A 9-bp deletion between COII/tRNA^{Lys} occurs in high frequency in eastern Asian and some African populations, because of its independent origins at different phylogenetic backgrounds (Soodyall et al. 1996). It was shown recently that some Indian populations also harbor the 9-bp deletion while clustering separately from Asian and African deleted lineages (Watkins et al. 1999). We found that 21% of Koyas and 3% of Chenchus harbored the deletion at the haplogroup M background. The Chenchu type (16184-16223-16256G-16362) has been previously observed at notable frequencies (44%) among Irulas, another tribe from Andhra Pradesh with australoid anthropological features (Watkins et al. 1999). The presence of the 9-bp deletion at the haplogroup M background was also observed among Kadars of Tamil Nadu and Kerala (Edwin et al. 2002). The HVS-I motif associated with the 9-bp deletion in Koyas has not been observed in previously published studies. Whether the Koya and Chenchu 9-bp deletion types stem from the same deletion event is difficult to judge. They differ by seven HVS-I mutations, suggesting either an ancient common root or independent origins of the deletion.

The haplogroup R lineages of the Koyas (31%) and Chenchus (1%) did not further subdivide into western Eurasian-specific (HV, U, TJ, and R1; Macaulay et al. 1999) or eastern Eurasian-specific branches (B and R9; Kivisild et al. 2002) and showed a coalescence time of 73,000 ± 20,900 years, which overlaps with the age es-

timate 0(o)0(uEint)]TJoJig70(Chenlj/F7 1 Tfi41(and)-365(showe65(tatio

Table 4**Compound Y-Chromosomal Haplotypes in Chenchus and Koyas**

HAPLOTYPE	CLADE ^a	NO. OF REPEATS AT LOCUS						CHENCHU	KOYA	MATCH ^b
		DYS019	DYS388	DYS390	DYS391	DYS392	DYS393			
1	C	15	13	23	10	11	12	1		
2	C	16	13	24	10	11	12	1		
3	F	15	13	21	11	11	14		1	
4	F	15	14	21	11	11	14		1	
5	F	16	13	21	10	12	14		1	
6	F	16	13	21	11	10	14		1	
7	F	16	13	21	11	11	13		1	
8	F	16	13	21	11	11	14		2	
9	F	16	14	21	11	11	14		1	
10	F	17	13	21	11	10	14		2	
11	F	17	13	21	11	11	13		1	
12	H1	13	12	23	11	11	12	1		
13	H1	14	12	21	9	11	12	1		
14	H1	14	12	22	10	11	12		2	
15	H1	14	13	22	10	11	13		1	
16	H1	14	13	22	11	11	12		1	
17	H1	15	12	21	9	11	12	7		
18	H1	15	12	21	10	11	12	2		
19	H1	15	12	21	11	11	12	1		
20	H1	15	12	22	10	11	12	3	11	*
21	H1	15	12	22	10	11	13		3	*
22	H1	15	13	21	11	11	12		1	
23	H1	15	13	22	10	11	13		2	
24	H1	15	13	22	11	11	12		1	
25	H1	16	12	22	10	11	13		1	
26	H1	16	13	22	10	11	12		2	
27	H2	16	12	21	10	11	13		4	*
28	J2*	14	15	23	10	11	13	1		
29	J2e	15	14	24	11	11	12	1		*
30	J2e	15	15	24	10	11	12	1		*
31	L	14	12	22	10	13	11	2		
32	L	14	12	22	10	14	11	4		*
33	R1a	15	12	24	11	11	12		1	
34	R1a	15	12	25	10	11	13	1		*
35	R1a	15	12	26	10	11	13	1		*
36	R1a	16	12	24	10	11	13	1		*
37	R1a	17	12	24	11	11	13	1		*
38	R1a	16	12	24	11	11	13	7		*
39	R1b	14	13	24	11	13	13	1		*
40	R2	14	12	22	10	10	14	1		
41	R2	14	12	23	10	10	14	1		
42	R2	14	13	22	10	10	14	1		
Total								41	41	

^a According to YCC nomenclature.^b Compared to 239 Indian caste samples.

in Middle Eastern populations to the west (table 3). Unclassified derivatives of the general Eurasian clade F were observed most frequently (27%) in the Koyas.

In comparison with caste groups (see fig. 3 and table 3), both tribal populations showed significantly ($P < .01$) higher frequencies of haplogroup H1. The characteristic M52 A→C transversion has also been described at relatively high frequencies in populations of Tamil Nadu, in southern India (Wells et al. 2001). Among the caste groups, its frequency is the lowest among Punjabis

in the northwest. Interestingly, more than one-third of Andhra Pradesh middle and lower caste Y chromosomes were defined as clade 1R in a previous study (Bamshad

Table 5

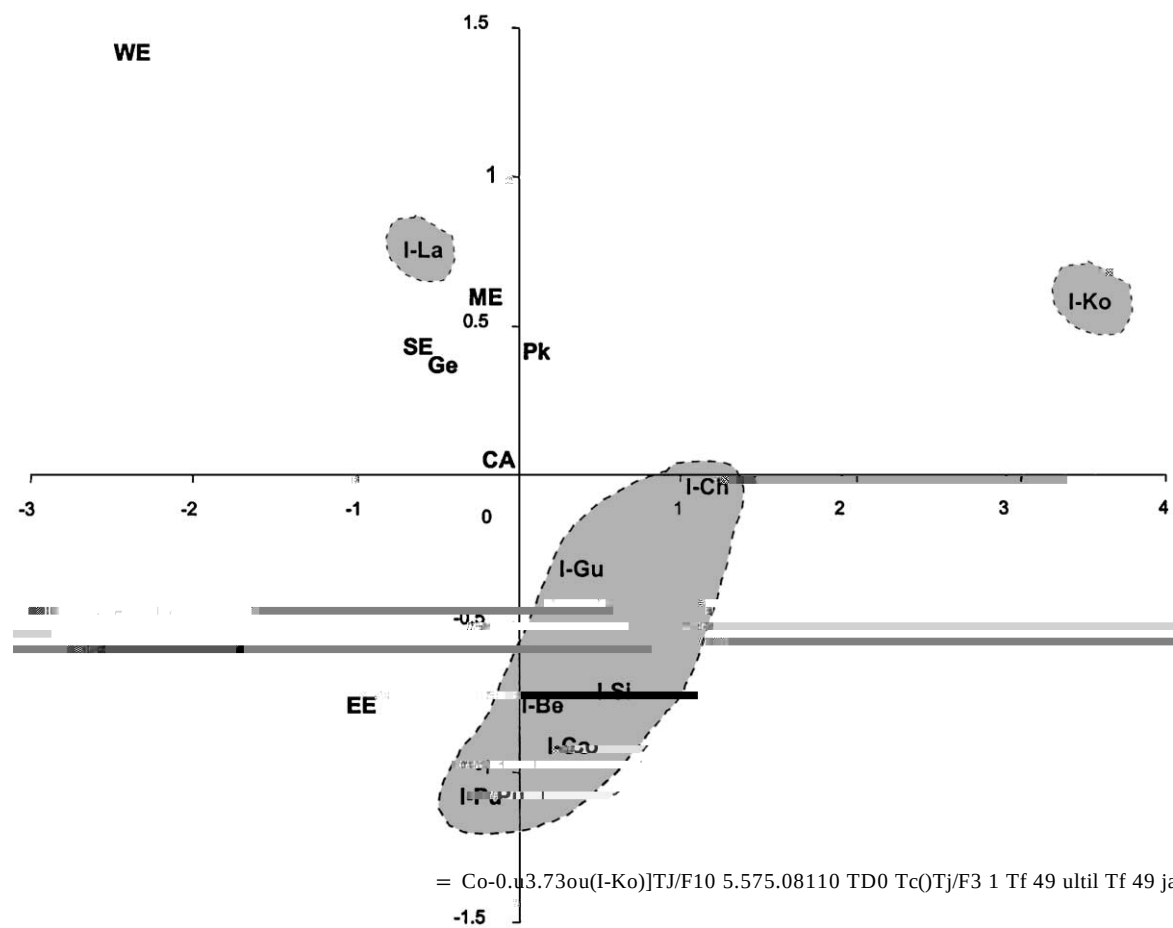


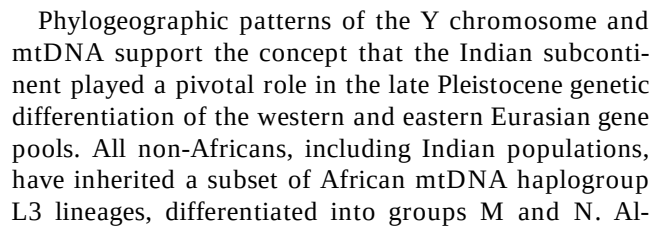
Figure 4 Multidimensional scaling plot of eight Indian and seven western Eurasian populations, using F_{st} distances calculated for 16 Y-chromosomal SNP haplogroups. From India: I-Ch = Chenchus, I-Ko

Table 6**MX1 Haplotypes of Chromosome 21 in Indian Populations, Compared with Continental Groups of the World**

POPULATION	NO. OF LINEAGES (95% CR FOR PROPORTION)									
	Ht1	Ht2	Ht3	Ht4	Ht5	Ht6	Ht7	Ht8	Ht9	Ht10
Chenchus (84) ^a	0 (.00-.04)	11 (.08-.22)	0 (.00-.04)	0 (.00-.04)	16 (.12-.29)	0 (.00-.04)	40 (.37-.58)	17 (.13-.30)	0 (.00-.04)	0 (.00-.04)
Koyas (56) ^a	0 (.00-.05)	4 (.03-.17)	0 (.00-.05)	0 (.00-.05)	15 (.17-.40)	0 (.00-.05)	16 (.18-.42)	21 (.26-.51)	0 (.00-.05)	0 (.00-.05)
Punjab (68) ^a	0 (.00-.04)	10 (.08-.25)	0 (.00-.04)	0 (.00-.04)	7 (.05-.20)	0 (.00-.04)	31 (.34-.57)	20 (.20-.41)	0 (.00-.04)	0 (.00-.04)
West Bengal (70) ^a	1 (.00-.08)	2 (.01-.10)	0 (.00-.04)	0 (.00-.04)	16 (.15-.34)	0 (.00-.04)	34 (.37-.60)	17 (.16-.36)	0 (.00-.04)	0 (.00-.04)
Pakistan (72) ^b	0 (.00-.04)	5 (.03-.15)	0 (.00-.04)	0 (.00-.04)	6 (.04-.17)	0 (.00-.04)	27 (.27-.49)	34 (.36-.59)	0 (.00-.04)	0 (.00-.04)
Anatolia (70) ^a	2 (.01-.10)	7 (.05-.19)	0 (.00-.04)	0 (.00-.04)	3 (.02-.12)	1 (.00-.08)	29 (.31-.53)	28 (.29-.52)	0 (.00-.04)	0 (.00-.04)
Europe (192) ^b	4 (.01-.05)	17 (.06-.14)	0 (.00-.02)	0 (.00-.02)	3 (.01-.05)	0 (.00-.02)	88 (.39-.53)	79 (.34-.48)	1 (.00-.03)	0 (.00-.02)
East Asia (118) ^b	0 (.00-.03)	3 (.01-.07)	0 (.00-.03)	0 (.00-.03)	59 (.41-.59)	11 (.05-.16)	45 (.30-.47)	0 (.00-.03)	0 (.00-.03)	0 (.00-.03)
Sub-Saharan Africa (102) ^b	9 (.05-.16)	3 (.01-.08)	0 (.00-.03)	0 (.00-.03)	22 (.15-.31)	23 (.16-.32)	33 (.24-.42)	9 (.05-.16)	0 (.00-.03)	3 (.01-.08)
Amerinds (120) ^b	0 (.00-.02)	14 (.07-.19)	0 (.00-.02)	0 (.00-.02)	78 (.56-.73)	0 (.00-.02)	2 (.01-.06)	26 (.15-.30)	0 (.00-.02)	0 (.00-.02)
Australia and PNG (76) ^b	2 (.01-.09)	10 (.07-.23)	16 (.13-.32)	2 (.01-.09)	27 (.26-.47)	0 (.00-.04)	15 (.12-.30)	4 (.02-.13)	0 (.00-.04)	0 (.00-.04)

^a From the present study.^b From Jin et al. 1999.

Discussion



- Female gene flow stratifies Hindu castes. *Nature* 395:651–652
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