# The Genetic Heritage of the Earliest Settlers Persists Both in Indian Tribal and Caste Populations

T. K,  $1^{1,7}$  S. R,  $1^{1}$  M. M, L,  $1^{1}$  S. ML, L,  $1^{2}$  K. KL,  $1^{1}$  J. PL,  $1^{1}$  E. M, L,  $1^{1}$  M, A, LL,  $1^{1}$  H.-. T,  $1^{1}$  S, L,  $3^{3}$  M. G,  $4^{4}$  E. L, L,  $5^{5}$  S. S. PL, L,  $6^{6}$  C, C,  $7^{7}$  R. K,  $7^{7}$  L. CL,  $1^{-5}$  S,  $1^{7}$  P. A.  $7^{7}$  L. R.  $7^{7}$  L. CL,  $1^{-5}$  S,  $1^{7}$  P. A.  $7^{7}$  L. R.  $7^{7}$  L. CL,  $1^{-5}$  S,  $1^{7}$  P. A.  $7^{7}$  L. R.  $7^{7}$  L. CL,  $1^{-5}$  S,  $1^{7}$  P. A.  $7^{7}$  L. R.  $7^{7}$  L. CL,  $1^{-5}$  S,  $1^{7}$  L,  $1^{6}$  C,  $1^{7}$  S,  $1^{7}$  L,  $1^{$ 

the Ida\_the Che ch adKa\_eeaa edf vaat 🛛 🕏 t ch da T t bag f 🛡 DNA & tDNA), the Y ch & e, a d e a t & a c a d e e c & a ed th f 🕐 ca te g dffeet at fIda, a e a thete adcetaAa.I the tDNA h geet caae, the Chech adK a cae ce at I da - ec c b a che f ha g Mad N that c ve fdffeet ca at a f 🛡 a ve the bc t e t. C a e ce ce t🛡 e gge t ea ate P e t ce e ett 🗗 e t f the A a a d gget that the e ha t beet ta e act e t f the e ette b ate t g at . H, L, a d R2 a e the 🖲 a Ida Y-ch 🗗 🗗 ahag that cc bth cate ad t ba at adae ae f d t de the bc t e t. Ha g R1a, ev a cated th the tat ve I d -A a va , a f d at the ghetfeec P abbta at a eative high feec (26%) the Chechtbe. The dig, t gethe th the hghe R1a-a cated h t ta date e eat d ve t I d a a d I a c a d th E e adcetaAa, gget that the adete Aat ght be the cefth hag. Hat e feeceftheMX1 c fch 🛡 🛡 e21 d t g hK a a dChech, a g th I da categ , ea a deate A a at . Ta e t gethe, the e e t h that I d a t ba a d ca te f 🛡 E de ve age f 🕏 the að ege et che tage f Pet ce e the ad et e A a ad have at eceved **v** tedge e f **v** e te a eg ce the H ce e. The h ge g a h f the **v** a **v** tDNA de gget that the e the A a Pet ceec a ta ette f 🕏 Af ca dhave a dY-ch 🛡 🖤 e f v ded the c af the be et d ffeet at f the d t ct ve eate a d e te E a a ge e

# Introduction

The origins of the culturally and genetically diverse pop-

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 (Roychoudhurv 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. Exogamous patrilocal 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 (Altais, 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).

# DAA \_\_\_

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. 1999*a*; Bamshad et al. 2001), 48 Haviks, 43 Mukris, and 7 Kadars 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, 2001*b*) 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.

#### 

Preparation of sequencing templates was performed according to methods described by Kaessmann et al. (1999). Purified products were sequenced with the DYEnamic 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  $\rho$ , 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. (2000*a*). In mtDNA coalescent calculations, using the estimator  $\rho$ , 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).

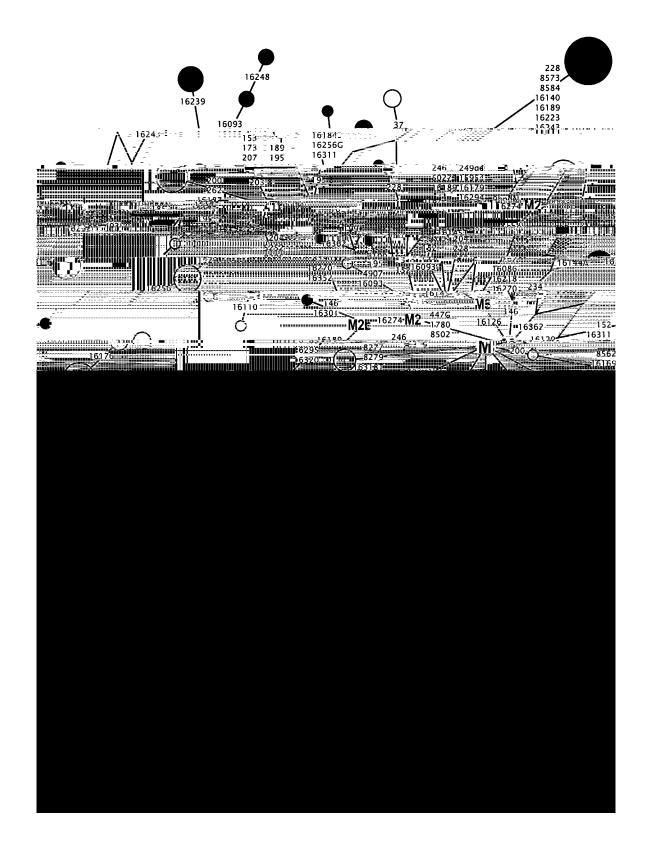
To calculate the 95% credible regions (CR85(W)r/F2-345-ucaoCR85(W,)]T485(trl20(wA30.grages)-u-475(po1aration)-41 Tc[(,)

							$1 \ 1 \ 1 \ 1 \ 1 \ 1 \ 1 \ 1 \ 1 \ 1 \$
			1				$egin{array}{cccccccccccccccccccccccccccccccccccc$
			6		6		$5 \ 5 \ 5 \ 8 \ 0 \ 5 \ 2 \ 8 \ 2 \ 3 \ 3 \ 3 \ 8 \ 4 \ 3 \ 4 \ 4 \ 7 \ 2 \ 7 \ 6 \ 9$
			5		4 6	1674-1880;	$3\ 7\ 8\ 2\ 2\ 9\ 4\ 2\ 3\ 6\ 9\ 9\ 7\ 6\ 0\ 0\ 0\ 0\ 5\ 6\ 0\ 2$
			1		4 3	4761-5260;	$7 \ 7 \ 4 \ 3 \ 5 \ 8 \ 9 \ 0 \ 7 \ 4 \ 4 \ 7 \ 1 \ 5 \ 8 \ 3 \ 6 \ 4 \ 9 \ 6 \ 6 \ 5$
Туре	Group	HVS-I (minus 16000)	9	HVS-II	7 e	8250-8710	а qааа fbgyecazugzowoua 1281609 TD0 Tca

 Table 1

 mtDNA Haplotypes in Chenchu and Koya Populations

25	M2a	223 270 274 292 319 352	C 73 204 263					+ +				1		*
26	M2a	223 270 274 319 352	C 73 204 263	G	1780 4769 5252 8396 8502 8701			+ + + -					2	**
27	M2a	223 270 274 319 352	C 73 203 204 263					+ +					1	**
28	M2a	93 223 243 248 270 319 352	C 73 195 204 263	G	1780 4769 5252 8396 8502 8701			+ + + -					2	
29	M2a	93 223 243 270 319 352	C 73 195 204 263	G	1780 4769 5252 8396 8502 8701			+ + + -					2	
30	M2b	167+C 183C 189 223 274 295 319 320	Т	G	1780 4769 8502 8701			+ + -	_				1	
31	M2b	183C 189 223 274 295 316 319 320	C 73 182 195 263	G	1780 4769 8502 8701			+ + -					1	
32	M2b	223 274 301 319	C 73 146 263	G	1780 4769 8502 8701		-	+ + + -					1	
33	M3	126 192 223	C 73 131 152 263	С	4769 8701	+ + +		+ + + -	+	-			2	**
34	M3	126 192 223	C 73 131 152 228 263					+ +					1	**
35	M3	126 223	C 73 131 152 263	С	4769 4935 8701	+		+ + + -					1	**
36	M3	126 223 362	C 73 131 152 263	С		+		+ + + -					1	
37	M3	93 126 145 223	T 73 195 263	С	4769 4907 8701	+		+ + + -				1		*
38	M6	188 223 231 362	C 73 146 263	С	4769 8701	+	+ –	+ + + -	+	- +	+	18		**
39	Ν	111 144 223 256 311		С –	-									



#### Table 2

#### Major mtDNA Lineage Clusters in India and Western Asia

POPULATION (n)	$D^{ m b}$	L1-L3	М	M2	M3	M5	M∆9bp	U2i, U7	U1, U3–U6, U*	HV <sup>c</sup> , TJ, N1, X	B, F <sup>d</sup>	R*
Tribal:												
Chenchus (96) <sup>e</sup>	.87	0 (.00–.03)	93 (.9199)	17 (.1127)	1 (.00–.06)	18 (.1228)	3 (.0109)	0 (.0003)	0 (.00–.03)	0 (.0003)	0 (.0003)	1 (.00–.06)
Koyas (81) <sup>e</sup>	.94	0 (.0004)	56 (.5878)	15 (.1228)	5 (.0314)	0 (.0004)	17 (.1431)	1 (.00–.07)	0 (.0004)	0 (.0004)	0 (.0004)	25 (.2242)
Tamil Nadu (49) <sup>f</sup>	.96	0 (.00–.06)	35 (.5882)	1 (.01–.11)	12 (.1538)	0 (.0006)	0 (.0006)	6 (.0624)	2 (.0114)	0 (.0006)	0 (.0006)	6 (.06–.24)
Western Bengal (34) <sup>f</sup>	.99	0 (.00–.08)	22 (.4879)	2 (.0219)	3 (.03–.23)	0 (.0008)	0 (.0008)	7 (.10–.37)	0 (.00–.08)	0 (.0008)	0 (.0008)	5 (.0730)
Caste:												
Western Bengalis (106) <sup>e</sup>	.97	0 (.00–.03)	76 (.63–.79)	4 (.02–.09)	7 (.03–.13)	6 (.0312)	0 (.00–.03)	10 (.05–.17)	1 (.01–.05)	6 (.0312)	0 (.00–.03)	12 (.0719)
Gujaratis and Konkanastha Br. (111) <sup>e</sup>	.99	0 (.00–.03)	53 (.39–.57)	5 (.0210)	7 (.03–.13)	0 (.0003)	0 (.00–.03)	20 (.1226)	5 (.0210)	11 (.06–.17)	5 (.0210)	12 (.06–.18)
Kerala/Karnataka (99) <sup>g</sup>	.96	0 (.00–.03)	63 (.5472)	15 (.0924)	6 (.03–.13)	15 (.0924)	0 (.0003)	20 (.1429)	1 (.01–.05)		0 (.00–.03)	9 (.05–.16)
Lambadis (86) <sup>h</sup>	.99	0 (.00–.03)	55 (.53–.73)	9 (.06–.19)	4 (.02–.11)	9 (.06–.19)	0 (.0003)	9 (.06–.19)	2 (.0108)	7 (.04–.16)	0 (.00–.03)	11 (.07–.22)
Lobanas (62) <sup>h</sup>	.98	0 (.00–.05)	34 (.43–.67)	3 (.02–.13)	3 (.02–.13)	5 (.04–.18)	0 (.0005)	2 (.0111)	1 (.00–.09)	5 (.04–.18)	0 (.00–.05)	11 (.10–.29)
Punjabis (112) <sup>e</sup>	.99	0 (.00–.03)	46 (.3250)	1 (.00–.05)	5 (.0210)	1 (.0005)	0 (.0003)	15 (.0821)	8 (.04–.14)	21 (.1327)	6 (.03–.11)	11 (.06–.17)
Sri Lanka (132) <sup>e</sup>	.99	0 (.00–.02)	77 (.50–.66)	9 (.04–.13)	6 (.02–.10)	2 (.0105)	0 (.00–.02)	19 (.09–.21)	5 (.0209)	11 (.05–.14)	2 (.0105)	18 (.09–.21)
Telugu, upper (59) <sup>i</sup>	.99	0 (.00–.05)	36 (.48–.72)	3 (.02–.14)	11 (.11–.30)	0 (.0005)	0 (.00–.05)	10 (.1029)	1 (.00–.09)	2 (.0111)	0 (.00–.05)	9 (.08–.27)
Telugu, middle (114) <sup>i</sup>	.99	0 (.00–.03)	73 (.55–.72)	7 (.03–.12)	4 (.01–.09)	5 (.0210)	0 (.00–.03)	10 (.05–.15)	1 (.01–.05)	6 (.03–.11)	0 (.00–.03)	24 (.15–.29)
Telugu, lower (70) <sup>i</sup>	.99	0 (.0004)	50 (.6081)	7 (.05–.19)	1 (.00–.08)	3 (.02–.12)	0 (.0004)	5 (.03–.16)	0 (.0004)	1 (.00–.08)	0 (.0004)	15 (.14–.32)
Uttar Pradesh (139) <sup>e,h</sup>	.99	0 (.00–.02)	79 (.49–.65)	4 (.01–.07)	14 (.06–.16)	0 (.0002)	0 (.00–.02)	21 (.1022)	3 (.01–.06)	9 (.04–.12)	2 (.0005)	20 (.1021)
West Asian:												
Iranians (440) <sup>e</sup>	.99	2 (.0002)	24 (.04–.08)	0 (.0001)	5 (.0103)	0 (.0001)	0 (.0002)	41 <sup>j</sup> (.07–.12)	90 (.1725)	245 (.5160)	2 (.0002)	17 (.0206)
Turks, Cappadocia (388) <sup>e</sup>	.99	1 (.00–.01)	16 (.03–.07)	0 (.0001)	0 (.0001)	0 (.0001)	0 (.0001)	7 (.01–.04)	93 (.20-29)	244 (.5868)	1 (.00–.01)	7 (.01–.04)
Middle East (406) <sup>e.k</sup>	.99	41 (.08–.13)	30 (.05–.10)	2 (.0002)	1 (.00–.01)	0 (.0001)	0 (.00–.03)	15 (.0206)	40 (.07–.13)	269 (.6271)	1 (.00–.01)	3 (.0002)

<sup>a</sup> 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.

<sup>b</sup> HVS-I haplotype diversity.

<sup>c</sup> Including pre-HV, as defined by Saillard et al. 2000*b*.
 <sup>d</sup> None of the Indian samples belonged to haplogroup B.

<sup>e</sup> From the present study.
 <sup>f</sup> From Roychoudhury et al. 2001.
 <sup>g</sup> From Moutain et al. 1995.

<sup>h</sup> From Kivisild et al. 1999.

<sup>i</sup> From Bamshad et al. 2001.

<sup>j</sup> Forty Iranians belonged to U7 and one belonged to U2i.

<sup>k</sup> The Middle East sample includes 202 Kuwaitis and 204 Saudi Arabians.

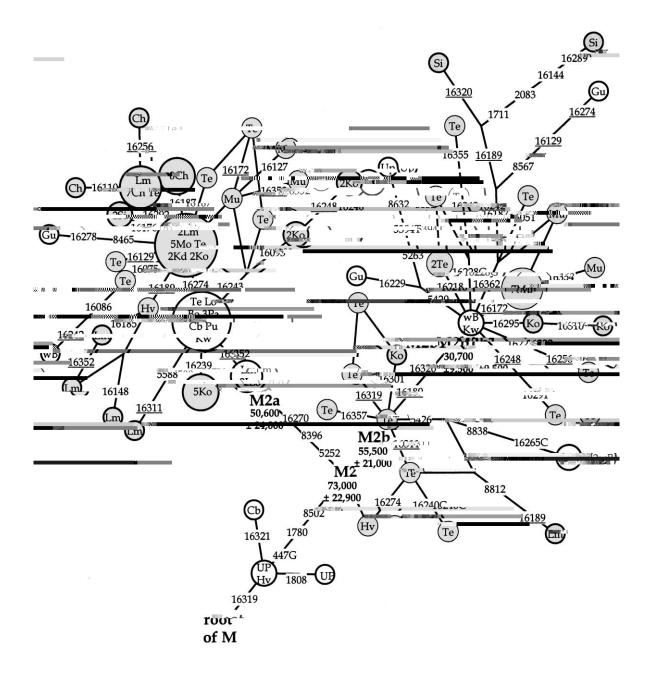


Figure 2 A network of haplogroup M2 haplotypes. Circle areas are proportional to haplotypes frequencies. Variant bases are numbered

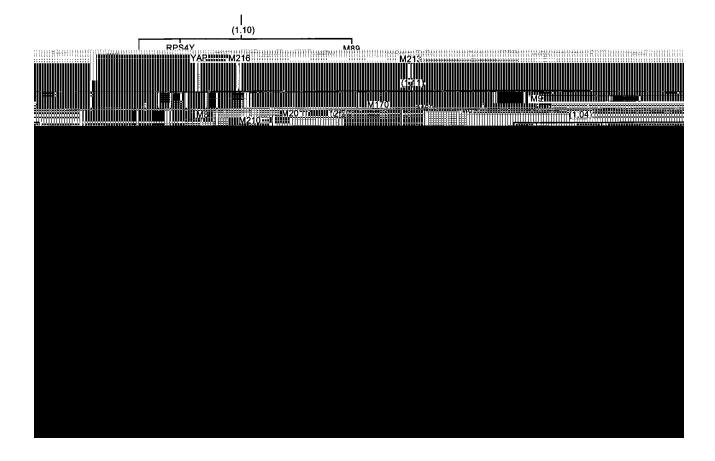


Figure 3 Y-chromosomal SNP tree and haplogroup frequencies in 8 Indian populations. Haplogroup defining markers (and their background average variances of 6 STR loci) are shown along the branches of the tree.

lable 3
---------

Major Y-Chromosomal Haplogroups in India Compared with Western Eurasia

Frequency (95% CR for Proportion)									
POPULATION ( <i>n</i> )	H1 (M52)	L (M11)	R2 (M124)	Reference					
India:									
Punjab (66)	.03 (.0110)	.12 (.0622)	.05 (.0213)	Present study					
West Bengal (31)	.10 (.0425)	.00 (.0009)	.23 (.1240)	Present study					
AP tribes (82)	.49 (.4364)	.07 (.04–.15)	.04 (.0110)	Present study					
AP tribes (67)	.10 (.0520)	.04 (.0212)		Ramana et al. 200					
AP castes (125)	.10 (.0616)	.08 (.04–.14)		Ramana et al. 200					
Tamil Nadu (259)	.17 (.1322)	.29 (.2435)	.07 (.0511)	Wells et al. 2001					
Tadjikistan (168)	.01 (.0004)	.11 (.0716)	.06 (.0311)	Wells et al. 2001					
Uzbeks (366)	.02 (.0104)	.03 (.0205)	.02 (.0104)	Wells et al. 2001					
Kyrgyzstan (92)	.00 (.0003)	.00 (.0003)	.02 (.0108)	Wells et al. 2001					
Kazakstan (95)	.01 (.0006)	.01 (.0006)	.01 (.0006)	Wells et al. 2001					
Iran (52)	.00 (.0006)	.04 (.0113)	.02 (.0110)	Wells et al. 2001					
Near East (101)	.01 (.0005)	.02 (.0107)	.00 (.0003)	Semino et al. 2000					
Caucasus (147)	.00 (.0002)	.01 (.0005)	.00 (.0002)	Wells et al. 2001					
Europe (839)	.00 (.0001)	.01 (.0001)	.00 (.0001)	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 *Mnl*I, 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<sup>Lys</sup> 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 9bp 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  $\pm$  20,900 years, which overlaps with the age estimate 0(o)0(uEint)]TJojig70(Chenlj/F7 1 Tfi41(and)-365(showe65(tatio

## Table 4

Compound Y-Chromosomal Haplotypes in Chenchus and Koyas

			N							
Haplotype	CLADE <sup>a</sup>	DYS019	DYS388	DYS390	DYS391	DYS392	DYS393	Chenchu	Koya	Match <sup>b</sup>
1	С	15	13	23	10	11	12	1		
2	С	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	12	24	11	13	13	1		*
40	R2	14	12	22	10	10	14	1		
40	R2	14	12	23	10	10	14	1		
42	R2	14	12	22	10	10	14	1		
Total	100		10	~~	10	10		$\frac{1}{41}$	41	

<sup>a</sup> According to YCC nomenclature.
 <sup>b</sup> 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

324

Table 5

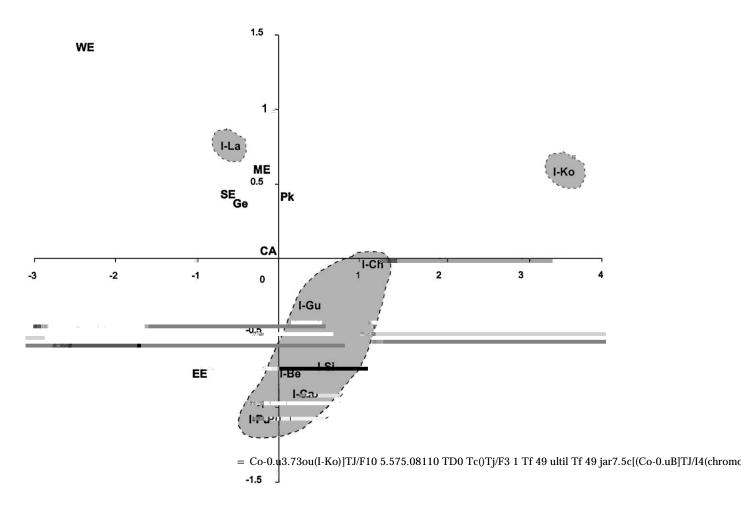


Figure 4 Multidimensional scaling plot of eight Indian and seven western Eurasian populations, using  $F_{st}$  distances calculated for 16 Y-chromosomal SNP haplogroups. From India: I-Ch = Chenchus, I-Ko

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

	NO. OF LINEAGES (95% CR FOR PROPORTION)											
POPULATION	Ht1	Ht2	Ht3	Ht4	Ht5	Ht6	Ht7	Ht8	Ht9	Ht10		
Chenchus (84) <sup>a</sup>	0 (.0004)	11 (.08–.22)	0 (.0004)	0 (.0004)	16 (.1229)	0 (.0004)	40 (.3758)	17 (.1330)	0 (.0004)	0 (.0004)		
Koyas (56) <sup>a</sup>	0 (.0005)	4 (.03–.17)	0 (.00–.05)	0 (.00–.05)	15 (.1740)	0 (.00–.05)	16 (.1842)	21 (.2651)	0 (.00–.05)	0 (.0005)		
Punjab (68) <sup>a</sup>	0 (.0004)	10 (.08–.25)	0 (.0004)	0 (.0004)	7 (.05–.20)	0 (.0004)	31 (.3457)	20 (.2041)	0 (.0004)	0 (.0004)		
West Bengal (70) <sup>a</sup>	1 (.0008)	2 (.0110)	0 (.0004)	0 (.0004)	16 (.1534)	0 (.0004)	34 (.3760)	17 (.1636)	0 (.0004)	0 (.0004)		
Pakistan (72) <sup>b</sup>	0 (.0004)	5 (.0315)	0 (.0004)	0 (.0004)	6 (.04–.17)	0 (.0004)	27 (.2749)	34 (.3659)	0 (.0004)	0 (.0004)		
Anatolia (70) <sup>a</sup>	2 (.0110)	7 (.05–.19)	0 (.0004)	0 (.0004)	3 (.0212)	1 (.00–.08)	29 (.3153)	28 (.2952)	0 (.0004)	0 (.0004)		
Europe (192) <sup>b</sup>	4 (.0105)	17 (.06–.14)	0 (.0002)	0 (.0002)	3 (.0105)	0 (.0002)	88 (.3953)	79 (.34–.48)	1 (.0003)	0 (.0002)		
East Asia (118) <sup>b</sup>	0 (.0003)	3 (.0107)	0 (.0003)	0 (.0003)	59 (.4159)	11 (.0516)	45 (.3047)	0 (.0003)	0 (.0003)	0 (.0003)		
Sub-Saharan Africa (102) <sup>b</sup>	9 (.0516)	3 (.0108)	0 (.0003)	0 (.0003)	22 (.1531)	23 (.1632)	33 (.2442)	9 (.0516)	0 (.0003)	3 (.0108)		
Amerinds (120) <sup>b</sup>	0 (.0002)	14 (.0719)	0 (.0002)	0 (.0002)	78 (.56-73)	0 (.0002)	2 (.0106)	26 (.1530)	0 (.0002)	0 (.0002)		
Australia and PNG (76) <sup>b</sup>	2 (.01–.09)	10 (.07–.23)	16 (.1332)	2 (.0109)	27 (.26-47)	0 (.0004)	15 (.1230)	4 (.02–.13)	0 (.00–.04)	0 (.0004)		

<sup>a</sup> From the present study. <sup>b</sup> From Jin et al. 1999.

dian populations considered here. Similarly, haplotype 8, which is common in Europe but absent in eastern Asia, was found in India at low frequencies. As is the case in other Eurasian and African populations, haplotypes 3 and 4, which are specific to Australian and Papuan populations, were not found in India. In contrast to the significant differences of haplotype frequencies that were observed between Indian and other world populations, none of the differences in haplotype frequencies was significant within India between caste and tribal groups.

### Discussion

Phylogeographic patterns of the Y chromosome and mtDNA support the concept that the Indian subcontinent played a pivotal role in the late Pleistocene genetic differentiation of the western and eastern Eurasian gene pools. All non-Africans, including Indian populations, have inherited a subset of African mtDNA haplogroup L3 lineages, differentiated into groups M and N. Al-

Kivisild et al.: Origins of Indian Castes and Tribes

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

- Bandelt H-J, Forster P, Röhl A (1999) Median-joining networks for inferring intraspecific phylogenies. Mol Biol Evol 16:37–48
- Bandelt H-J, Forster P, Sykes BC, Richards MB (1995) Mitochondrial portraits of human populations using median networks. Genetics 141:743–753
- Bergen AW, Wang CY, Tsai J, Jefferson K, Dey C, Smith KD, Park SC, Tsai S-J, Goldman D (1999) An Asian–Native American paternal lineage identified by RPS4Y resequencing and by microsatellite haplotyping. Ann Hum Genet 63: 63–80
- Bertorelle G, Excoffier L (1998) Inferring admixture proportions from molecular data. Mol Biol Evol 15:1298–1311
- Bhattacharyya NP, Basu P, Das M, Pramanik S, Banerjee R, Roy B, Roychoudhury S, Majumder PP (1999) Negligible male gene flow across ethnic boundaries in India, revealed by analysis of Y-chromosomal DNA polymorphisms. Genome Res 9:711–719
- Bhowmick PK (1992) The Chenchus of the forests and plateaux: Calcutta. Institute of Anthropology, Calcutta
- Cavalli-Sforza LL, Menozzi P, Piazza A (1994) The history and geography of human genes. Princeton University Press, Princeton
- Chikhi L, Bruford MW, Beaumont MA (2001) Estimation of admixture proportions: a likelihood-based approach using Markov chain Monte Carlo. Genetics 158:1347–1362
- Chikhi L, Nichols RA, Barbujani G, Beaumont MA (2002) Y genetic data support the Neolithic demic diffusion model. Proc Natl Acad Sci USA 99:11008–11013
- Cruciani F, Santolamazza P, Shen P, Macaulay V, Moral P, Olckers A, Modiano D, Holmes S, Destro-Bisol G, Coia V, Wallace DC, Oefner PJ, Torroni A, Cavalli-Sforza LL, Scozzari R, Underhill PA (2002) A back migration from Asia to sub-Saharan Africa is supported by high-resolution analysis of human Y-chromosome haplotypes. Am J Hum Genet 70: 1197–1214
- Edwin D, Vishwanathan H, Roy S, Usha Rani M, Majumder P (2002) Mitochondrial DNA diversity among five tribal populations of southern India. Curr Sci 83:158–163
- Forster P, Harding R, Torroni A, Bandelt H-J (1996) Origin and evolution of Native American mtDNA variation: a reappraisal. Am J Hum Genet 59:935–945
- Forster P, Torroni A, Renfrew C, Röhl A (2001) Phylogenetic star contraction applied to Asian and Papuan mtDNA evolution. Mol Biol Evol 18:1864–1881
- Hammer MF, Redd AJ, Wood ET, Bonner MR, Jarjanazi H, Karafet T, Santachiara-Benerecetti S, Oppenheim A, Jobling MA, Jenkins T, Ostrer H, Bonne-Tamir B (2000) Jewish and Middle Eastern non-Jewish populations share a common pool of Y-chromosome biallelic haplotypes. Proc Natl Acad Sci USA 97:6769–6774
- Helgason A, Sigurðadóttir S, Gulcher J, Ward R, Stefanson K (2000*a*) mtDNA and the origins of the Icelanders: deciphering signals of recent population history. Am J Hum Genet 66:999–1016
- Helgason A, Sigurðardóttir S, Nicholson J, Sykes B, Hill EW, Bradley DG, Bosnes V, Gulcher JR, Ward R, Stefánsson K

(2000*b*) Estimating Scandinavian and Gaelic ancestry in the male settlers of Iceland. Am J Hum Genet 67:697–717

- Herrnstadt C, Elson JL, Fahy E, Preston G, Turnbull DM, Anderson C, Ghosh SS, Olefsky JM, Beal MF, Davis RE, Howell N (2002) Reduced-median-network analysis of complete mitochondrial DNA coding-region sequences for the major African, Asian, and European haplogroups. Am J Hum Genet 70:1152–1171
- Ingman M, Kaessmann H, Pääbo S, Gyllensten U (2000) Mitochondrial genome variation and the origin of modern humans. Nature 408:708–713
- Jin L, Underhill PA, Doctor V, Davis RW, Shen P, Cavalli-Sforza LL, Oefner PJ (1999) Distribution of haplotypes from a chromosome 21 region distinguishes multiple prehistoric human migrations. Proc Natl Acad Sci USA 96:3796–3800
- Kaessmann H, Heissig F, von Haeseler A, Pääbo S (1999) DNA sequence variation in a non-coding region of low recombination on the human X chromosome. Nat Genet 22:78–81
- Kalaydjieva L, Calafell F, Jobling MA, Angelicheva D, de Knijff P, Rosser ZH, Hurles ME, Underhill P, Tournev I, Marushiakova E, Popov V (2001) Patterns of inter- and intragroup genetic diversity in the Vlax Roma as revealed by Y chromosome and mitochondrial DNA lineages. Eur J Hum Genet 9:97–104
- Karafet T, Xu L, Du R, Wang W, Feng S, Wells RS, Redd AJ, Zegura SL, Hammer MF (2001) Paternal population history of East Asia: sources, patterns, and microevolutionary processes. Am J Hum Genet 69:615–628
- Karafet TM, Zegura SL, Posukh O, Osipova L, Bergen A, Long J, Goldman D, Klitz W, Harihara S, deKnijff P, Wiebe V, Griffiths RC, Templeton AR, Hammer MF (1999) Ancestral Asian source(s) of new world Y-chromosome founder haplotypes. Am J Hum Genet 64:817–831
- Kayser M, Brauer S, Weiss G, Schiefenhovel W, Underhill PA, Stoneking M (2001) Independent histories of human Y chromosomes from Melanesia and Australia. Am J Hum Genet 68:173–190
- Kayser M, Brauer S, Weiss G, Underhill PA, Roewer L, Schiefenhovel W, Stoneking M (2000) Melanesian origin of Polynesian Y chromosomes. Curr Biol 10:1237–1246
- Ke Y, Su B, Song X, Lu D, Chen L, Li H, Qi C, Marzuki S, Deka R, Underhill P, Xiao C, Shriver M, Lell J, Wallace D, Wells RS, Seielstad M, Oefner P, Zhu D, Jin J, Huang W, Chakraborty R, Chen Z, Jin L (2001) African origin of modern humans in East Asia: a tale of 12,000 Y chromosomes. Science 292:1151–1153
- Kivisild T, Bamshad MJ, Kaldma K, Metspalu M, Metspalu E, Reidla M, Laos S, Parik J, Watkins WS, Dixon ME, Papiha SS, Mastana SS, Mir MR, Ferak V, Villems R (1999a) Deep common ancestry of Indian and western-Eurasian mitochondrial DNA lineages. Curr Biol 9:1331–1334
- Kivisild T, Kaldma K, Metspalu M, Parik J, Papiha SS, Villems R (1999*b*) The place of the Indian mitochondrial DNA variants in the global network of maternal lineages and the peopling of the Old World. In: Deka R, Papiha SS (eds) Genomic diversity. Kluwer/Academic/Plenum Publishers, New York, pp 135–152
- Kivisild T, Papiha SS, Rootsi S, Parik J, Kaldma K, Reidla M, Laos S, Metspalu M, Pielberg G, Adojaan M, Metspalu E,

Mastana SS, Wang Y, Gölge M, Demirtas H, Schnakenberg E, De Stefano GF, Geberhiwot T, Claustres M, Villems R (2000) An Indian ancestry: a key for understanding human diversity in Europe and beyond. In: Renfrew C, Boyle K (eds) Archaeogenetics: DNA and the population prehistory of Europe. McDonald Institute for Archaeological Research, University of Cambridge, Cambridge, pp 267–279

- Kivisild T, Tolk H-V, Parik J, Wang Y, Papiha SS, Bandelt H-J, Villems R (2002) The emerging limbs and twigs of the east Asian mtDNA tree. Mol Biol Evol 19:1737–1751
- Maca-Meyer N, Gonzalez AM, Larruga JM, Flores C, Cabrera VM (2001) Major genomic mitochondrial lineages delineate early human expansions. BMC Genet 2:13
- Macaulay VA, Richards MB, Hickey E, Vega E, Cruciani F, Guida V, Scozzari R, Bonné-Tamir B, Sykes B, Torroni A (1999) The emerging tree of west Eurasian mtDNAs: a synthesis of control-region sequences and RFLPs. Am J Hum Genet 64:232–249
- Majumder PP (2001) Indian caste origins: genomic insights and future outlook. Genome Res 11:931–932
- Majumder P, Dey B (2001) Absence of the HIV-1 protective Delta ccr5 allele in most ethnic populations of India. Eur J Hum Genet 9:794-796
- Mountain JL, Hebert JM, Bhattacharyya S, Underhill PA, Ottolenghi C, Gadgil M, Cavalli-Sforza LL (1995) Demographic history of India and mtDNA-sequence diversity. Am J Hum Genet 56:979–992
- Nebel A, Filon D, Brinkmann B, Majumder PP, Faerman M, Oppenheim A (2001) The Y chromosome pool of Jews as part of the genetic landscape of the Middle East. Am J Hum Genet 69:1095–1112
- Nebel A, Landau-Tasseron E, Filon D, Oppenheim A, Faerman M (2002) Genetic evidence for the expansion of Arabian tribes into the Southern Levant and North Africa. Am J Hum Genet 70:1594–1596
- Nei M, Roychoudhury A (1993) Evolutionary relationships of human populations on a global scale. Mol Biol Evol 10: 927–943
- Papiha SS (1996) Genetic variation in India. Hum Biol 68: 607–628
- Passarino G, Semino O, Magri C, Al-Zahery N, Benuzzi G, Quintana-Murci L, Andellnovic S, Bullc-Jakus F, Liu A, Arslan A, Santachiara-Benerecetti AS (2001) The 49a,f haplotype 11 is a new marker of the EU19 lineage that traces migrations from northern regions of the Black Sea. Hum Immunol 62:922–932
- Passarino G, Semino O, Modiano G, Bernini LF, Santachiara Benerecetti AS (1996) mtDNA provides the first known marker distinguishing proto-Indians from the other Caucasoids; it probably predates the diversification between Indians and Orientals. Ann Hum Biol 23:121–126
- Qamar R, Ayub Q, Mohyuddin A, Helgason A, Mazhar K, Mansoor A, Zerjal T, Tyler-Smith C, Mehdi SQ (2002) Ychromosomal DNA variation in Pakistan. Am J Hum Genet 70:1107–1124
- Quintana-Murci L, Krausz C, Zerjal T, Sayar SH, Hammer MF, Mehdi SQ, Ayub Q, Qamar R, Mohyuddin A, Radhakrishna U, Jobling MA, Tyler-Smith C, McElreavey K (2001) Y-chromosome lineages trace diffusion of people and

languages in southwestern Asia. Am J Hum Genet 68: 537–542

- Quintana-Murci L, Semino O, Bandelt H-J, Passarino G, McElreavey K, Santachiara-Benerecetti AS (1999) Genetic evidence of an early exit of *Homo sapiens sapiens* from Africa through eastern Africa. Nat Genet 23:437–441
- Raitio M, Lindroos K, Laukkanen M, Pastinen T, Sistonen P, Sajantila A, Syvanen A (2001) Y-chromosomal SNPs in Finno-Ugric-speaking populations analyzed by minisequencing on microarrays. Genome Res 11:471–482
- Ramana GV, Su B, Jin L, Singh L, Wang N, Underhill P, Chakraborty R (2001) Y-chromosome SNP haplotypes suggest evidence of gene flow among caste, tribe, and the migrant Siddi populations of Andhra Pradesh, South India. Eur J Hum Genet 9:695–700
- Redd A, Roberts-Thomson J, Karafet T, Bamshad M, Jorde L, Naidu J, Walsh B, Hammer MF (2002) Gene flow from the Indian subcontinent to Australia: evidence from the Y chromosome. Curr Biol 12:673–677
- Richards M, Macaulay V, Hickey E, Vega E, Sykes B, Guida V, Rengo C, et al (2000) Tracing European founder lineages in the near eastern mtDNA pool. Am J Hum Genet 67: 1251–1276
- Rosser ZH, Zerjal T, Hurles ME, Adojaan M, Alavantic D, Amorim A, Amos W, et al (2000) Y-chromosomal diversity

- Shouse B (2001) Archaeology: spreading the word, scattering the seeds. Science 294:988–989
- Shriver MD, Smith MW, Jin L, Marcini A, Akey JM, Deka R, Ferrell RE (1997) Ethnic-affiliation estimation by use of population-specific DNA markers. Am J Hum Genet 60:957– 964
- Singh KS (1997) The scheduled tribes. In: Singh KS (ed) People of India. Vol III. Oxford University Press, Oxford
- Soodyall H, Vigilant L, Hill AV, Stoneking M, Jenkins T (1996) mtDNA control-region sequence variation suggests multiple independent origins of an "Asian-specific" 9-bp deletion in sub-Saharan Africans. Am J Hum Genet 58:595–608
- Stringer C (2000) Coasting out of Africa. Nature 405:24-25
- Su B, Xiao C, Deka R, Seielstad MT, Kangwanpong D, Xiao J, Lu D, Underhill P, Cavalli-Sforza LL, Chakraborty R, Jin L (2000) Y chromosome haplotypes reveal prehistorical migrations to the Himalayas. Hum Genet 107:582–590
- Su B, Xiao J, Underhill P, Deka R, Zhang W, Akey J, Huang W, Shen D, Lu D, Luo J, Chu J, Tan J, Shen P, Davis R, Cavalli-Sforza L, Chakraborty R, Xiong M, Du R, Oefner P, Chen Z, Jin L (1999) Y-chromosome evidence for a northward migration of modern humans into Eastern Asia during the last ice age. Am J Hum Genet 65:1718–1724
- Tarn W (1951) The Greeks in Bactria & India. Cambridge University Press, Cambridge
- Thangaraj K, Ramana GV, Singh L (1999) Y-chromosome and mitochondrial DNA polymorphisms in Indian populations. Electrophoresis 20:1743–1747
- Thurin M (1999) The Chenchu of the Indian Deccan. In: Lee RB, Daily R (eds) The Cambridge encyclopedia of hunters and gatherers. Cambridge University Press, Cambridge, pp 252–256
- Underhill PA, Jin L, Lin AA, Mehdi SQ, Jenkins T, Vollrath D, Davis RW, Cavalli-Sforza LL, Oefner PJ (1997) Detection of numerous Y chromosome biallelic polymorphisms by denaturing high-performance liquid chromatography. Genome Res 7:996–1005
- Underhill PA, Passarino G, Lin AA, Marzuki S, Oefner PJ, Cavalli-Sforza LL, Chambers GK (2001*a*) Maori origins, Ychromosome haplotypes and implications for human history in the Pacific. Hum Mutat 17:271–280
- Underhill PA, Passarino G, Lin AA, Shen P, Mirazon Lahr M, Foley R, Oefner PJ, Cavalli-Sforza LL (2001*a*) The phylogeography of Y chromosome binary haplotypes and the or-

igins of modern human populations. Ann Hum Genet 65: 43–62

- Underhill PA, Shen P, Lin AA, Jin L, Passarino G, Yang WH, Kauffman E, Bonné-Tamir B, Bertranpetit J, Francalacci P, Ibrahim M, Jenkins T, Kidd JR, Mehdi SQ, Seielstad MT, Wells RS, Piazza A, Davis RW, Feldman MW, Cavalli-Sforza LL, Oefner PJ (2000) Y chromosome sequence variation and the history of human populations. Nat Genet 26:358–361
- Walter H, Danker-Hopfe H, Bhasin MK (1991) Anthropologie Indiens. Gustav Fischer Verlag, Stuttgart
- Watkins WS, Bamshad M, Dixon ME, Bhaskara Rao B, Naidu JM, Reddy PG, Prasad BV, Das PK, Reddy PC, Gai PB, Bhanu A, Kusuma YS, Lum JK, Fischer P, Jorde LB (1999) Multiple origins of the mtDNA 9-bp deletion in populations of South India. Am J Phys Anthropol 109:147–158
- Weale ME, Yepiskoposyan L, Jager RF, Hovhannisyan N, Khudoyan A, Burbage-Hall O, Bradman N, Thomas MG (2001) Armenian Y chromosome haplotypes reveal strong regional structure within a single ethno-national group. Hum Genet 109:659–674
- Wells RS, Yuldasheva N, Ruzibakiev R, Underhill PA, Evseeva I, Blue-Smith J, Jin L, et al (2001) The Eurasian heartland: a continental perspective on Y-chromosome diversity. Proc Natl Acad Sci USA 98:10244–10249
- Wilson JF, Weiss DA, Richards M, Thomas MG, Bradman N, Goldstein DB (2001) Genetic evidence for different male and female roles during cultural transitions in the British Isles. Proc Natl Acad Sci USA 98:5078–5083
- Y Chromosome Consortium, The (2002) A nomenclature system for the tree of human Y-chromosomal binary haplogroups. Genome Res 12:339–348
- Zerjal T, Pandya A, Santos FR, Adhikari R, Tarazona E, Kayser M, Evgafov O, Singh L, Thangaray K, Destro-Bisol G, Thomas MG, Qamar R, Mehdi SQ, Rosser ZH, Hurles ME, Jobling MA, Tyler-Smith C (1999) The use of Y-chromosomal DNA variation to investigate population history: recent male spread in Asia and Europe. In: Papiha S, Deka R, Chakraborty R (eds) Genomic diversity: applications in human population genetics. Kluwer Academic/Plenum Publishers, New York, pp 91–101
- Zerjal T, Wells R, Yuldasheva N, Ruzibakiev R, Tyler-Smith C (2002) A genetic landscape reshaped by recent events: Ychromosomal insights into central Asia. Am J Hum Genet 71:466–482