

## FOR THE RECORD

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# Allele Frequencies of 20 Y-Chromosomal Short Tandem Repeat Loci in a Tribal Population of Deccan Plateau, India

**POPULATION:** Eighty male individuals from a nomadic tribal population belonging to Dravidian and Indo-Caucasian ethnicities from Deccan Plateau, Andhra Pradesh, India (1), were analyzed in the present study.

**KEYWORDS:** forensic science, DNA typing, population genetics, Y-chromosome, short tandem repeats, DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I/II, YCA II, DYS426, DYS388, DYS439, DYS437, GATA H4, DYS460, DYS447, DYS438, DYS448, allele frequencies, human identification, male contamination, Deccan Plateau, Andhra Pradesh, India

The allele frequency distribution at 20 Y-chromosomal short tandem repeat (STR) loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385I/II, YCA II, DYS426, DYS388, DYS439, DYS437, GATA H4, DYS460, DYS447, DYS438, and DYS448) was analyzed in a nomadic tribal population of Andhra Pradesh, India. Peripheral blood samples were obtained by venipuncture from 80 consenting, healthy, unrelated male individuals. Genomic DNA was extracted from blood using the standard phenol–chloroform extraction method (2). Co-amplification of 20 Y-STRs was carried out using fluorescent-labeled primers in a 10  $\mu$ L final reaction volume containing 10 ng of genomic DNA, as validated in the laboratory (Sahoo and Kashyap, personal communication). All amplifications were performed on a GeneAmp PCR 9700 thermal cycler (Applied Biosystems, Foster City, CA). Amplified PCR fragments were separated on 5% polyacrylamide denaturing gel using an ABI 377 Automated DNA Sequencer with GS500 LIZ<sup>TM</sup> used as an internal lane standard. Sizing of the products was performed using the ABI 377 GeneScan Analysis software version (3.7). Alleles were designated according to the number of repeats (3–6).

Allele and haplotype frequencies were estimated by the gene count method. Gene and haplotype diversities for the 20 Y-STRs were calculated according to Nei's formula (7),

$$h = n \left( 1 - \sum p_i^2 \right) / (n - 1)$$

where  $n$  represents the number of chromosomes sampled and  $p_i$  represents the frequency of the  $i$ th allele or haplotype, using the software ARLEQUIN ver 2.0 software package (8). The discriminatory capacity of the 20 Y-STRs was calculated by dividing the number of different haplotypes seen in a given population by the total number of individuals in the population.

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The haplotype data of the selected 20 Y-chromosomal loci among the tribal population of Andhra Pradesh are presented in Table 1. The results of the allele frequency are shown in Tables 2–4. A total of 77 unique haplotypes were observed in 80 male individuals. In our population, samples collected from a defined geographical area, we obtained a haplotype diversity discrimination index of  $0.9625 \pm 0.0013$ . Tetranucleotide marker DYS385 (a and b) was found to be the most polymorphic (0.942) and DYS391 as the least polymorphic loci (0.460). The combined gene diversity of the studied 20 Y-STRs was calculated as 0.998702. On comparison with the U.S. Y-STR haplotype reference database (9) using the minimal set of minimal haplotype markers, no match was found with the haplotypes in the present study. Similarly, no equivalent haplotype matches were observed with the European or Asian databases (10,11). Thus, the present study shows that 20 Y-STR haplotype systems are highly discriminatory and polymorphic.

A comparative analysis was performed using the Y-STR loci mentioned in SWGDAM and the European Forensic Community (minimal and extended haplotype) and the 20 Y-STRs used in the current study, and noteworthy results obtained are shown in Table 5. Characterization of further highly informative Y-STRs will increase the power of exclusion of Y-chromosomal haplotypes and increase the usefulness of these markers in human identification and paternity analysis. The data generated certainly augment the available Y-STR information for Indian populations (12,13).

The complete data set is available on request through electronic mail from the corresponding author.

### Acknowledgments

The authors are grateful to the participants and Tribal Welfare Department, Government of Andhra Pradesh, and the various nongovernmental organizations for facilitating in this objective. This work was carried out through a grant under X Plan Project of CFSL, Kolkata. The useful suggestions and assistance of Ms.

TABLE 1—*Y-chromosomal haplotypes based on 20 Y-chromosomal STR loci in a tribal population of Deccan Plateau, India.*

Haplotype	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385	DYS 438	DYS 439	YCA II	GATA H4	DYS 388	DYS 426	DYS 437	DYS 447	DYS 448	DYS 460	nFrequency
HT_01	15	12	28	22	10	10	11	10,13	9	16	17,19	12	11	11	14	23	22	10	1 0.0125
HT_02	15	12	28	22	10	10	12	13,16	9	16	17,19	12	12	11	14	23	22	10	1 0.0125
HT_03	15	12	28	22	10	10	12	10,13	9	16	18,19	12	12	11	14	23	22	10	1 0.0125
HT_04	15	12	28	22	10	10	11	13,14	9	16	18,19	12	12	11	14	23	22	10	1 0.0125
HT_05	15	12	28	22	9	10	11	12,14	9	16	18,19	12	11	11	14	23	22	10	1 0.0125
HT_06	15	12	28	22	13	10	11	13,14	9	16	18,19	12	11	12	14	23	22	10	1 0.0125
HT_07	15	12	28	22	10	10	11	13,14	9	16	18,19	12	11	11	14	24	21	10	1 0.0125
HT_08	15	14	30	21	9	10	13	13,15	9	15	18,19	12	12	11	14	23	22	10	1 0.0125
HT_09	13	13	30	22	10	11	13	11,15	9	16	18	12	12	11	14	22	22	10	1 0.0125
HT_10	15	12	28	22	9	10	11	13,14	9	16	18,19	12	10	11	14	23	22	10	1 0.0125
HT_11	15	12	28	22	10	10	11	13,14	9	16	18,19	12	11	11	14	24	22	10	1 0.0125
HT_12	15	12	28	22	10	10	12	13,14	9	16	17,19	12	11	11	14	24	22	10	1 0.0125
HT_13	15	12	28	22	10	11	12	13,14	9	16	17,19	12	11	11	14	24	22	10	1 0.0125
HT_14	15	12	28	22	10	11	12	13,14	9	16	17,19	12	11	11	14	23	22	10	1 0.0125
HT_15	15	12	28	22	10	11	12	13,14	9	21	17,19	12	11	11	14	23	22	10	1 0.0125
HT_16	15	13	29	22	10	10	12	13,16	9	21	17,19	12	13	11	14	23	21	10	1 0.0125
HT_17	15	12	28	22	10	10	12	12,14	9	16	17,19	12	11	11	14	23	22	10	1 0.0125
HT_18	16	12	28	22	9	10	11	13,15	9	16	17,19	11	13	11	14	23	22	10	1 0.0125
HT_19	13	13	30	21	10	14	13	11,16	10	18	17,18	12	13	11	15	22	22	10	1 0.0125
HT_20	15	12	28	23	10	10	12	13,15	9	17	18,19	12	11	11	14	24	21	10	1 0.0125
HT_21	14	12	29	22	10	14	10	10,14	10	18	17	12	12	11	14	23	22	10	1 0.0125
HT_22	15	14	31	25	10	11	12	8,11	11	18	17,20	12	12	11	13	25	23	11	1 0.0125
HT_23	14	15	31	21	9	12	12	8,9	12	16	14,17	10	12	12	13	25	22	9	1 0.0125
HT_24	15	13	30	24	10	10	11	11,15	9	17	17,18	10	10	11	14	27	22	10	1 0.0125
HT_25	15	14	31	25	10	12	12	8,11	11	18	17,20	13	11	12	13	25	23	11	2 0.025
HT_26	15	14	31	25	12	11	12	8,11	11	18	17,20	13	12	12	13	25	23	11	1 0.0125
HT_27	14	14	31	21	12	11	12	8,9	12	18	14,17	10	12	11	14	25	22	9	1 0.0125
HT_28	14	14	30	21	10	10	12	9,13	11	17	14,17	10	11	11	14	25	22	9	1 0.0125
HT_29	14	15	31	21	10	10	12	9,13	11	17	14,17	10	12	11	14	25	21	9	1 0.0125
HT_30	15	13	30	24	10	11	11	11,14	12	17	18	10	14	11	14	27	22	10	1 0.0125
HT_31	14	15	30	21	10	10	12	9,13	11	17	14,17	10	11	11	14	25	22	9	1 0.0125
HT_32	15	14	31	25	10	12	12	8,11	11	18	17,20	13	11	11	13	25	23	11	1 0.0125
HT_33	15	14	31	25	10	11	12	8,11	11	18	17,20	13	11	11	13	25	23	11	1 0.0125
HT_34	15	13	30	24	10	10	11	11,15	9	18	17,18	13	13	11	14	27	22	10	1 0.0125
HT_35	14	14	28	21	10	10	12	13,22	11	18	12,17	13	12	11	14	25	22	9	1 0.0125
HT_36	14	14	30	21	12	10	12	9,13	11	16	14,17	10	12	11	14	25	22	9	1 0.0125
HT_37	16	12	28	22	11	11	11	12,15	9	17	17,19	13	12	11	13	22	22	9	1 0.0125
HT_38	16	12	28	22	11	10	11	12,14	9	16	17,18	13	12	11	13	25	22	9	1 0.0125
HT_39	14	13	29	24	10	10	13	8,11	11	17	16,19	11	12	11	14	25	23	9	1 0.0125
HT_40	14	13	29	24	11	12	11	9,11	11	17	17,21	13	12	12	14	25	22	11	1 0.0125
HT_41	15	14	30	24	11	12	11	9,11	11	17	17,21	13	11	11	14	25	22	11	1 0.0125
HT_42	15	14	30	24	11	12	11	9,11	11	17	18,21	13	14	11	14	25	22	10	1 0.0125
HT_43	15	15	32	23	11	10	12	9,12	11	17	17,22	13	12	12	13	26	23	11	1 0.0125
HT_44	15	10	27	24	10	10	12	11,12	9	18	19	12	13	11	13	?	24	11	1 0.0125
HT_45	15	13	31	25	10	10	12	9,12	11	18	17,21	13	16	11	13	24	23	10	1 0.0125
HT_46	14	13	29	22	10	14	11	8,15	10	17	16,19	11	12	11	15	26	23	11	1 0.0125
HT_47	16	13	31	25	11	10	12	9,12	11	19	17,21	13	11	12	13	24	23	10	1 0.0125
HT_48	14	12	28	22	11	13	10	10,15	10	18	17,19	11	11	11	14	24	22	11	1 0.0125
HT_49	14	14	30	24	11	12	11	9,11	11	17	17,21	13	14	11	14	25	22	11	1 0.0125
HT_50	14	13	29	24	11	12	11	9,11	11	17	17,21	13	13	12	14	25	22	12	1 0.0125
HT_51	14	14	31	25	10	12	12	11,16	11	18	16,20	11	12	12	14	26	21	10	1 0.0125
HT_52	16	13	31	25	11	10	12	9,12	11	18	17,21	13	12	12	13	24	23	11	1 0.0125
HT_53	14	13	29	24	11	12	11	9,11	11	17	17,21	14	13	12	13	25	22	11	1 0.0125
HT_54	14	15	31	23	10	11	13	10,11	11	16	17,19	11	12	12	15	26	22	11	1 0.0125
HT_55	14	14	31	25	10	12	12	11,15	11	18	17,20	11	12	12	14	27	21	10	1 0.0125
HT_56	14	14	31	25	10	12	12	11,16	11	18	18,21	11	12	12	14	26	21	10	1 0.0125
HT_57	14	13	29	24	11	12	11	9,11	11	18	18,22	13	13	12	14	24	22	11	1 0.0125
HT_58	15	13	29	22	10	10	11	14,23	8	15	18,20	11	12	11	13	24	22	11	1 0.0125
HT_59	14	13	29	24	9	15	12	11,15	11	16	18,20	11	12	12	13	28	25	10	1 0.0125
HT_60	14	13	30	22	10	10	12	13,19	9	16	17,21	11	16	11	14	27	23	12	1 0.0125
HT_61	14	13	31	25	10	10	12	14,18	9	16	17,21	11	16	11	14	26	24	11	1 0.0125
HT_62	14	13	30	23	10	10	12	13,19	9	16	17,21	11	12	11	14	27	23	12	3 0.0375
HT_63	16	13	29	23	10	9	13	11,17	11	17	17,20	12	12	12	15	26	22	10	1 0.0125
HT_64	16	13	30	23	10	10	12	13,19	9	16	17,21	11	12	11	14	26	23	11	1 0.0125
HT_65	16	13	29	23	10	9	13	11,17	11	17	17,20	12	13	12	14	26	22	10	1 0.0125
HT_66	15	14	30	22	10	10	11	14,15	10	16	17,20	12	12	11	13	22	22	10	1 0.0125
HT_67	15	14	30	22	10	10	11	14,15	10	16	17,20	12	11	11	13	22	22	10	1 0.0125
HT_68	15	14	30	22	10	10	11	14,15	10	16	17,20	12	13	11	13	22	22	10	1 0.0125
HT_69	14	13	31	23	10	10	12	13,19	9	16	17,21	11	12	11	14	27	23	11	1 0.0125
HT_70	15	13	31	25	10	10	12	10,13	11	18	17,22	13	12	12	13	23	23	12	1 0.0125
HT_71	14	13	31	23	10	10	12	14,18	9	16	17,21	11	12	11	14	26	24	11	1 0.0125

TABLE 1—Continued.

Haplotype	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385	DYS 438	DYS 439	YCA II	GATA H4	DYS 388	DYS 426	DYS 437	DYS 447	DYS 448	DYS 460	nFrequency
HT_72	15	14	29	23	10	9	13	11,18	11	16	17,20	12	12	12	15	26	22	10	1 0.0125
HT_73	16	13	31	25	10	10	12	9,13	11	15	17,22	13	12	12	13	23	23	11	1 0.0125
HT_74	15	13	30	25	10	10	12	10,12	11	15	17,22	13	12	12	13	23	23	11	1 0.0125
HT_75	15	14	30	22	10	10	11	14,15	10	16	17,20	12	12	12	13	22	22	10	1 0.0125
HT_76	14	12	28	22	10	13	10	10,15	10	15	17,24	12	12	12	14	22	22	11	1 0.0125
HT_77	14	13	30	23	10	9	13	11,16	11	17	17,20	12	12	12	15	26	22	10	1 0.0125
Haplotype diversity																			0.998702

n, no. of individuals; STR, short tandem repeat.

TABLE 2—Allele frequencies and gene diversities at 16 Y-STR loci in a tribal population of Deccan Plateau, India.

Allele	DYS 438	DYS 389II	DYS 439	DYS 437	DYS 389I	DYS 391	DYS 390	DYS 393	DYS 426	DYS 392	DYS 19	DYS 388	GATA H4	DYS 460	DYS 448	DYS 447
6																
7																
8	0.013															
9	0.400					0.075				0.050					0.125	
10	0.112				0.013	<b>0.712</b>		0.038		0.138		0.025	0.100	<b>0.488</b>		
11	<b>0.438</b>					0.162		0.325	<b>0.675</b>	0.175		0.262	0.225	0.312		
12	0.038				0.262	0.038		<b>0.525</b>	0.325	0.175		<b>0.512</b>	<b>0.387</b>	0.075		
13				0.300	<b>0.387</b>	0.013		0.112		0.025	0.025	0.125	0.275			
14				<b>0.625</b>	0.275					0.038	0.375	0.038	0.013			
15			0.062	0.075	0.062					0.013	<b>0.488</b>					
16			<b>0.412</b>							0.112	0.038					
17			0.237													
18			0.250													
19			0.013													
20																
21			0.025				0.112								0.087	
22						<b>0.363</b>								<b>0.600</b>	0.100	
23						0.162								0.262	0.213	
24						0.162								0.038	0.138	
25						0.200								0.013	<b>0.262</b>	
26																0.150
27		0.013														0.112
28		0.262														0.013
29		0.162														
30		<b>0.287</b>														
31		0.262														
32		0.013														
33																
34																
<b>D</b>	0.634	0.754	0.707	0.514	0.702	0.460	0.763	0.605	0.439	0.630	0.608	0.650	0.714	0.643	0.562	0.822

The most frequent allele is given in bold numbers.  
D, gene diversity; STR, short tandem repeat.

TABLE 3—Allele frequencies and gene diversity at microsatellite locus *DYS385* (a and b) in a tribal population of Deccan Plateau, India.

Allele Combination Observed	Frequency	Allele Combination Observed	Frequency	Allele Combination Observed	Frequency
8,9	0.0250	10,14	0.0125	12,15	0.0125
8,11	0.0875	10,15	0.0250	13,14	<b>0.1125</b>
8,15	0.0125	11,12	0.0125	13,15	0.0375
9,11	0.0875	11,14	0.0125	13,16	0.0250
9,12	0.0500	11,15	0.0625	13,19	0.0750
9,13	0.0625	11,16	0.0500	13,22	0.0125
10,11	0.0125	11,17	0.0250	14,15	0.0500
10,12	0.0125	11,18	0.0125	14,18	0.0250
10,13	0.0375	12,14	0.0375	14,23	0.0125
<b>D</b>	0.9420				

The most frequent allele is given in bold numbers.  
D, gene diversity.

TABLE 4—Allele frequencies and gene diversity at microsatellite locus YCA II (a and b) in a tribal population of Deccan Plateau, India.

Allele Combination Observed	Frequency	Allele Combination Observed	Frequency	Allele Combination Observed	Frequency
12,17	0.0125	17,19	0.1500	18,19	0.1125
14,17	0.0750	17,20	0.1875	18,20	0.0250
16,19	0.0250	17,21	<b>0.2000</b>	18,21	0.0250
16,20	0.0125	17,22	0.0500	18,22	0.0125
17	0.0125	17,24	0.0125	19	0.0125
17,18	0.0500	18	0.0250		
<b>D</b>	0.8760				

The most frequent allele is given in bold numbers.  
D, gene diversity.

TABLE 5—Comparative analysis of a combination of Y-STR loci used in forensic evaluation for a tribal population, Deccan Plateau, India.

Parameters	20 Y-STRs (in the Present Study)	SWGDM Haplotype	Extended Haplotype	Minimal Haplotype
No. of individuals	80	80	80	80
No. of haplotypes	77	64	63	60
Random match probabilities	0.001298	0.007278	0.007900	0.009500
Haplotype diversity	0.998702	0.992720	0.992100	0.990500
Discriminatory power	0.9625	0.8	0.7875	0.75
Most frequent haplotype and frequency	3 0.038	4 0.05	4 0.05	4 0.05

STR, short tandem repeat.

Sanghamitra Sahoo are highly acknowledged. G. H. B. was assisted by a fellowship from Council of Scientific and Industrial Research (CSIR), New Delhi, India. Special thanks are due to Prof. Leonor Gusmao, Porto, Portugal, for providing the HapYDiv Software for analysis.

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