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Y chromosomal STRs haplotypes in two populations from Bolivia

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Abstract

In the present study, we typed our previously reported two microsatellite markers, DXYS241 and DXYS266 together with a basic set of nine Y-STRs (DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DXYS156Y, DYS413) on Y chromosomes from two Bolivian populations. Unrelated males from communities living at high- ($N = 59$) and low- ($N = 142$) altitude, were studied. Combining the alleles into 11 Y-STRs haplotypes revealed that the high-altitude population is significantly less diverse than the low-altitude population. Haplotype diversities of 0.927 ± 0.029 and 0.996 ± 0.002 were found within the high-altitude, and the low-altitude populations, respectively. Within the high-altitude population 40 haplotypes were detected, whereas in the low-altitude population 113 haplotypes were found. Only three haplotypes were shared between both populations.

Haplotyping-based discrimination using the 11 Y-STRs including our new two microsatellite markers DXYS241 and DXYS266 was shown to be powerful than using the conventional 9 Y-STRs, especially for the low-altitude Bolivian population.

This 11 Y-STRs-based haplotyping system shows a very high potential for discrimination and could provide an ideal tool for forensic analysis and population studies. Moreover, this study includes data about two Bolivian populations which were not previously reported, this will help in building a world-wide database for future use in forensic and legal studies.

Keywords

Y chromosome; Microsatellite; Y-STRs; Haplotypes; Bolivian populations

Population

Blood samples from 59 high-altitude Bolivian males and 142 low-altitude Bolivian males were collected from healthy blood donors. The subjects in the high-altitude group were from typical rural communities of the Andean Altiplano, living at an approximate elevation of 4000–4100 m above sea level, and located 280 km southwest from the city of La Paz. Details of that community were presented elsewhere [1,2]. The subjects from low-altitude were from the Alto Beni, subtropical Amazonian lowland colonies at an elevation of ~300 m located about 200 km southeast of La Paz along Alto Beni River, an upper tributary of the Amazon River. Details of those subjects were presented previously [3].

Extraction

Genomic DNA was extracted from the collected blood samples using the QIAamp Blood Kit from Qiagen (Hilden, Germany).

PCR

The 11 Y-STR loci (DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DXYS156Y, DYS413, DXYS241 and DXYS266) were amplified by the polymerase chain reaction (PCR) using four multiplex systems (system I: DYS19, DYS389I/II; system II: DYS390, DYS391, DYS392; system III: DYS393, DXYS156Y, YCAIII; system IV: DXYS241 and DXYS266). For primers and PCR conditions see Kayser et al. [4] and Lee et al. [5].

Typing

For analysis, 2 µl of the PCR products were mixed with 3 µl of formamide buffer and run on denaturing polyacrylamide gels on an ABI 377 sequencer using a TAMRA 500 internal size standard in each lane. Raw genotype data were analyzed using GENESCAN software (ABI). Allele nomenclature follows Kayser et al. [6] and de Knijff et al. [7].

Data analysis

Haplotype and allele frequencies were calculated by the counting method. Estimation of haplotype and gene diversities together with intra- and inter-population indices and analysis of molecular variance (AMOVA) were computed using ARLEQUIN version 2.000, <http://igb.unige.ch/arlequin>, developed by Schneider, Roessli and Excoffier [8].

Results

See Table 1

Other remarks

We analyzed Bolivian male samples from high- and low-altitudes using our previously reported haplotyping system, which is based on a single multiplex PCR for three polymorphic microsatellite markers, DXYS241, DXYS265 and DXYS266 [5]. Then, we compared the frequencies of the different alleles of these markers in the two Bolivian populations with our previous data for Japanese, Caucasian-American and African-American populations. All populations exhibited alleles 227 bp (CA₁₄) and 162 bp (CA₁₂) of DXYS241 and DXYS265, respectively, as the predominant alleles. However, at DXYS266, the 216 bp (CA₁₈) and 218 bp (CA₁₉) alleles were prevalent within the Bolivian populations, but were not found in other populations. We studied the haplotype diversity in both Bolivian populations using a basic set of nine Y-STRs (DYS19, DYS389I/II, DYS390,

DYS391, DYS392, DYS393, DXYS156Y, DYS413) only. At this stage of the study we did not combine our data of the three newly developed markers of Lee et al. [5]. When using only the 9 Y-STRs, the results showed a haplotype diversity of 0.91 ± 0.032 and 0.98 ± 0.003 in the low- and high-altitude populations, respectively.

We eliminated the marker DXYS265 from the haplotyping system because of its low diversity in Bolivian population.

Thus, we used haplotypes comprising the above two microsatellite markers (DXYS241 and DXYS266) together with the basic set of the nine Y-STRs (DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DXYS156Y, DYS413), for further studying the genetic diversity within and between the two Bolivian populations. We could detect 150 different haplotypes in both the high- and low-altitude populations. Only three haplotypes were shared between both populations. Within the high-altitude population 40 haplotypes were detected whereas, in the low-altitude population 113 haplotypes were found (Table 1). Haplotype diversities of 0.927 ± 0.029 and 0.996 ± 0.002 were found within the high-altitude, and the low-altitude populations, respectively. This haplotype diversity indicates that the high-altitude population is significantly less diverse than the low-altitude population. Interestingly, haplotype H22, which is present in both populations, exhibited the highest frequency among all haplotypes in the high-altitude population, 27.1% (16/59), while only 2.1% (3/142) of the low-altitude males have this haplotype (H22). However, still the Y-chromosomal haplotypes can differentiate effectively within and between the two Bolivian populations. We conclude that combining our two new microsatellite markers DXYS241 and DXYS266 with the other 9 Y-STRs within a standard haplotyping system improves the power of discrimination, and could provide an ideal tool for forensic analysis and population studies. These results can present a part of the genetic portrait of the peopling of Americas to discover the genetic diversity, genetic distance and genetic structure with estimation of the timing of migration waves of the New World founders and inhabitants [9]. Moreover, this study includes data about two Bolivian populations which were not previously reported, this will help in building a world-wide database for future use in forensic and legal studies.

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Table 1

Y chromosome haplotypes constructed by combinations of the typing results of 11 Y-STRs among the two, high- and low-altitude Bolivian populations

H	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DXYSI56Y	DYS413	DXYSI241	DXYSI266	High	Low
H1	13	10	25	23	10	17	13	2	23	14	19	1	0
H2	13	10	26	25	11	16	12	2	21	14	18	1	0
H3	13	10	26	25	11	16	12	2	24	14	18	1	0
H4	13	10	26	24	11	16	12	2	21	14	18	1	0
H5	13	11	27	23	10	16	13	2	23	14	18	1	0
H6	13	11	27	23	10	17	13	2	23	14	19	1	0
H7	13	11	27	23	10	17	14	2	23	14	19	1	0
H8	13	11	27	24	10	15	12	2	23	14	18	1	0
H9	13	11	28	23	10	15	12	2	23	13	18	1	0
H10	13	11	28	23	10	17	14	2	23	14	19	1	0
H11	13	11	28	20	10	16	12	2	23	14	18	1	0
H12	13	11	28	21	10	15	12	2	24	14	18	1	0
H13	13	11	28	20	10	15	12	2	24	14	18	1	0
H14	13	11	29	24	10	15	12	2	25	14	18	1	0
H15	13	11	30	22	11	15	12	2	23	14	18	1	0
H16	13	11	30	23	10	17	13	2	23	14	19	1	0
H17	13	12	27	23	9	15	12	2	22	14	18	1	0
H18	13	12	28	22	10	18	14	2	22	14	19	1	0
H19	13	12	28	25	10	15	12	2	23	12	18	1	0
H20	13	12	28	22	10	18	13	2	22	14	19	2	0
H21	13	12	28	22	10	17	14	2	23	14	18	1	0
H22	13	12	29	23	10	17	13	2	23	14	19	16	3
H23	13	12	29	22	10	18	13	2	22	14	19	1	0
H24	13	12	29	25	9	15	12	2	22	14	18	1	0
H25	13	12	29	23	10	17	13	2	23	14	18	1	1
H26	13	12	29	23	11	16	12	2	23	13	18	1	0
H27	13	12	29	26	11	16	12	2	24	14	18	1	0
H28	13	12	29	23	10	14	13	2	23	14	19	1	3
H29	13	12	29	23	10	16	13	2	23	14	19	1	0

H	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DXYS156Y	DYS413	DXYS241	DXYS266	High	Low
H30	13	12	29	24	10	17	13	2	23	14	19	1	0
H31	13	12	29	24	10	15	12	2	22	14	18	2	0
H32	14	10	27	26	11	16	12	2	24	14	18	1	0
H33	14	10	28	24	10	15	12	2	25	14	18	1	0
H34	14	11	27	23	11	16	12	2	24	14	19	1	0
H35	14	11	27	23	10	17	14	2	23	14	19	1	0
H36	14	11	27	25	10	14	12	2	23	14	16	2	0
H37	14	11	27	23	11	15	12	2	23	14	16	1	0
H38	14	11	27	24	11	14	12	2	23	14	18	1	0
H39	14	11	27	26	11	16	12	2	24	14	18	1	0
H40	14	11	27	24	11	16	12	2	24	14	18	2	0
H41	13	12	28	23	10	15	12	2	23	13	18	0	4
H42	14	11	28	25	11	14	12	2	23	14	18	0	1
H43	14	11	28	24	11	14	12	2	23	14	18	0	1
H44	14	9	26	24	10	14	12	2	22	14	18	0	1
H45	14	9	26	24	10	15	12	2	22	14	18	0	1
H46	14	9	26	24	10	15	12	2	25	14	18	0	1
H47	14	11	26	24	10	15	12	2	20	14	16	0	1
H48	14	11	26	24	10	14	12	2	20	13	18	0	1
H49	13	11	27	23	10	15	12	2	23	14	18	0	1
H50	14	10	26	24	11	14	12	2	23	14	17	0	3
H51	14	10	26	23	11	14	12	2	23	14	17	0	1
H52	14	10	26	24	11	14	12	2	23	13	17	0	1
H53	13	11	27	24	12	15	12	2	24	14	18	0	2
H54	14	11	26	24	11	14	12	2	20	14	16	0	1
H55	14	11	26	24	10	14	12	2	20	14	16	0	1
H56	13	11	26	23	10	14	12	2	23	14	18	0	1
H57	14	10	27	24	10	17	12	2	23	14	18	0	1
H58	14	10	26	24	10	15	12	2	20	14	16	0	1
H59	14	11	25	24	10	14	12	2	22	14	16	0	1
H60	15	11	28	24	10	14	12	2	20	14	16	0	1

H	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DXYS156Y	DYS413	DXYS241	DXYS266	High	Low
H61	13	12	28	23	10	15	12	2	23	14	18	0	4
H62	15	11	28	24	10	13	12	2	21	15	16	0	1
H63	13	11	28	23	10	13	12	2	23	13	19	0	1
H64	14	11	26	24	10	17	12	2	20	14	16	0	1
H65	16	10	27	22	10	14	11	2	17	15	16	0	1
H66	13	10	27	24	10	12	12	2	22	14	18	0	1
H67	14	11	27	27	11	15	12	2	23	13	16	0	1
H68	13	11	27	23	10	15	12	2	25	14	18	0	1
H69	14	10	27	24	11	15	12	2	23	14	17	0	1
H70	13	12	29	23	10	17	13	2	23	13	19	0	2
H71	13	11	28	24	10	14	13	2	22	13	18	0	1
H72	14	10	26	24	11	16	12	2	23	14	17	0	1
H73	13	11	27	23	10	15	12	2	23	13	18	0	2
H74	13	9	27	24	10	15	13	2	25	14	18	0	2
H75	13	11	28	24	10	15	12	2	23	13	18	0	1
H76	14	10	26	24	11	14	12	2	20	14	17	0	1
H77	13	9	27	24	10	15	13	2	23	14	18	0	2
H78	13	9	27	24	10	15	13	2	24	13	18	0	1
H79	13	10	26	24	10	15	13	2	23	14	18	0	1
H80	13	10	27	24	10	15	12	2	23	13	18	0	1
H81	13	10	27	24	10	15	12	2	25	14	18	0	1
H82	13	10	27	24	10	15	12	2	22	14	18	0	1
H83	13	11	28	23	10	15	12	2	25	14	18	0	2
H84	13	11	28	23	10	17	13	2	23	14	19	0	1
H85	13	11	27	23	11	17	12	2	23	13	19	0	1
H86	13	11	28	23	10	15	12	2	22	13	18	0	1
H87	13	12	29	24	10	15	12	2	23	12	18	0	1
H88	13	12	29	27	10	16	12	2	23	14	16	0	1
H89	13	12	29	23	10	16	12	2	25	14	18	0	1
H90	13	12	29	23	10	15	12	2	25	14	18	0	4
H91	13	12	28	23	10	18	13	2	23	14	19	0	1

H	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DXYS156Y	DYS413	DXYS241	DXYS266	High	Low
H92	13	12	28	24	10	15	12	2	22	13	18	0	1
H93	13	11	29	24	10	15	12	2	24	14	18	0	1
H94	13	11	28	24	10	15	12	2	22	14	17	0	2
H95	13	12	29	23	10	15	12	2	25	13	18	0	2
H96	14	10	28	24	11	14	12	2	23	14	16	0	1
H97	13	11	28	24	10	15	12	2	23	13	17	0	1
H98	14	11	28	27	11	14	12	2	23	15	18	0	1
H99	14	11	28	27	11	14	12	2	23	14	16	0	1
H100	13	11	28	23	10	17	13	2	23	13	18	0	1
H101	13	11	28	23	10	17	13	2	23	13	19	0	1
H102	13	10	27	24	10	16	12	2	23	13	18	0	1
H103	13	10	27	24	10	15	12	2	22	12	17	0	1
H104	14	11	28	24	11	14	12	2	23	13	16	0	1
H105	13	12	28	23	10	15	12	2	23	14	17	0	1
H106	13	12	28	23	10	15	13	2	23	14	18	0	1
H107	13	12	29	23	10	15	13	2	23	13	18	0	1
H108	13	12	29	23	10	17	13	2	23	13	18	0	1
H109	14	10	27	24	10	15	12	2	22	13	18	0	2
H110	13	11	27	23	11	16	12	2	22	12	17	0	1
H111	13	11	28	24	11	17	13	2	23	14	17	0	1
H112	14	11	28	24	11	19	12	2	23	13	16	0	1
H113	13	12	29	23	10	19	13	2	23	13	18	0	1
H114	17	11	28	21	10	14	14	1	20	13	16	0	1
H115	13	11	28	24	11	16	12	2	24	14	18	0	1
H116	13	12	28	23	10	17	13	2	23	13	19	0	1
H117	13	11	28	24	11	16	12	2	24	14	17	0	1
H118	13	12	29	23	10	19	13	2	23	14	19	0	2
H119	13	12	29	23	10	18	13	2	23	14	19	0	1
H120	14	10	27	24	10	15	12	2	22	14	18	0	1
H121	13	11	28	24	9	14	12	2	21	13	16	0	1
H122	13	12	31	24	9	18	12	2	23	14	18	0	1

H	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DXYS156Y	DYS413	DXYS241	DXYS266	High	Low
H123	13	12	29	23	10	19	13	2	25	14	19	0	1
H124	13	12	31	24	9	19	12	2	23	14	18	0	1
H125	13	12	29	23	10	19	13	2	23	13	19	0	1
H126	13	11	28	24	10	17	12	2	22	14	18	0	1
H127	14	10	26	24	11	16	12	2	23	14	16	0	1
H128	13	10	27	24	10	14	12	2	22	13	18	0	1
H129	13	12	29	23	10	18	12	2	23	14	19	0	1
H130	13	11	28	24	11	17	13	2	24	14	18	0	1
H131	13	12	30	23	10	17	12	2	23	14	18	0	1
H132	13	12	29	24	11	19	13	2	22	14	19	0	1
H133	13	12	29	23	11	19	13	2	23	14	19	0	3
H134	17	10	26	23	10	14	12	2	23	13	16	0	1
H135	13	12	29	23	10	20	13	2	21	13	19	0	1
H136	14	11	27	25	11	16	12	2	23	13	16	0	1
H137	14	11	27	25	11	16	12	2	23	14	16	0	1
H138	13	11	28	24	11	18	12	2	22	13	18	0	1
H139	13	10	27	25	10	17	12	2	23	14	16	0	1
H140	13	10	27	25	10	15	12	2	23	14	16	0	1
H141	13	11	28	24	10	15	11	2	22	14	18	0	1
H142	13	10	27	24	10	14	12	2	22	14	18	0	1
H143	13	12	29	23	10	15	12	2	23	14	18	0	1
H144	13	12	29	24	10	15	13	2	23	14	18	0	1
H145	13	12	28	23	10	15	12	2	25	14	18	0	1
H146	13	11	27	24	11	14	12	2	23	14	18	0	1
H147	13	9	26	24	10	15	13	2	23	14	18	0	1
H148	13	12	27	24	10	15	12	2	23	14	18	0	1
H149	13	12	28	23	10	17	13	2	23	14	18	0	3
H150	15	9	25	23	10	11	13	2	20	14	18	0	1

H22, H25 and H28 written in bold indicate the shared haplotypes between the high- and low-altitude Bolivian populations. H, Haplotype.