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Seven SNPs and YAP demonstrate that the chromosome Y lineages of Basques are different from Georgians and Berbers

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1. Introduction

The Y chromosome is an important tool for discriminating related males and populations. The short tandem repeat (STR) markers of the Y chromosome are highly variable and can therefore discriminate between chromosomes of different paternal lineages, which is of great usefulness in forensic casework.

The power of discrimination of each single nucleotide polymorphism (SNP) marker is lower than that of STR markers, although a broad combination of them also provides high discrimination capacity. SNP markers provide an additional advantage over STRs because they have more capacity for distinguishing human population groups and within specific populations. Thus, they are a useful tool for evaluating similarities between Basque, Georgian and Berber populations. The relationships found to date among these populations are based, principally, on linguistic data [1,4,6]. Previous genetic data have only related Basque and Berber populations [2].

Given the evidence reported so far, a more detailed study appeared necessary to evaluate the genetic relationships between Basque, Georgian and Berber populations. Accordingly, we decided to use SNP markers of the Y chromosome as the best tool for discriminating among populations.

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2. Materials and methods

Using the phenol-chloroform method, DNA was extracted from peripheral blood of 166 unrelated males from three different regions: 59 autochthonous Basques (34 from Arratia, 25 from Goiherri), 40 residents in the Basque Country, 49 from Georgia (12 residents, 32 autochthonous from Swanetia) and 18 Berbers from Mauritania.

Eight biallelic markers were studied (Table 1) and 30 ng of each sample was amplified in a final volume of 25 µl with 0.2 mM dNTPs, 1 µM primers, 1.5 mM $MgCl_2$ and 0.5 U Taq polymerase. Primers and PCR conditions are shown in Table 1. The PCR amplified products were digested with 0.1 U of restriction enzymes as indicated in Table 1.

Phylogenetic relationships were constructed in the form of median-joining networks of the haplotypes by using the Network 2.0 program. Neighbor-joining trees were also constructed from linearised matrix of genetic distance of Reynolds with the Phylip 3.4 package. The SPSS 9.0 program was used to make a Hierarchic Cluster analysis and a Multidimensional Scaling analysis based on the c2 distance. The allele frequencies for SNPs and YAP of worldwide reference populations have been published in Rosser et al. [7] and Karafet et al. [5] as follows: (1) Algerian, (2) Altaic, (3) Armenian, (4) Australian Aboriginal people, (5) Basques, (6) Basques of Arratia, (7) Basques of Goiherri, (8) Bavarian, (9) Belarusian, (10) Belgian, (11) Berber, (12) British, (13) Bulgarian, (14) Buryat, (15) Cheyenne, (16) Chuvash, (17) Cornish, (18) Czech, (19) Danish, (20) Dutch, (21) East African, (22) East Anglian, (23) East Bantus, (24) Egyptian, (25) Estonian, (26) Even, (27) Finns, (28) Forest Nentsi, (29) French, (30) Gambian, (31) Georgian, (32) German, (33) Gotlander, (34) Hungarian, (35) Icelandic, (36) India, (37) Indonesian, (38)

Table 1
Amplification parameters and enzymes used for the digestion of amplified PCR products

Marker	Restriction enzymes	Primers (5′–3′)	PCR conditions
YAP	YAP+: 455 bp YAP − : 150 bp	cag ggg aag ata aag aaa ta act gct aaa agg gga tgg at	94 °C/30 s; 54 °C/30 s; 72 °C/30 s, 33 cycles
Sy81	209 bp: *Nla*III 144 bp (G)/102 bp (A)	agg cac tgg tca gaa tga ag aat gga aaa tac agc tcc cc	94 °C/30 s; 60 °C/60 s; 72 °C/60 s, 32 cycles
Tat	112 bp: *Nla*III/*Hsp*II (T) *Mae*II(C) 85 bp/27 bp	gac tct gag tgt aga ctt gtg a gaa ggt gcc gta aaa gtg tga a	94 °C/30 s; 60 °C/30 s; 72 °C/30 s, 33 cycles
SRY-1532 (SRY-10831)	167 bp: *Dra*III 112 bp/55 bp	tcc tta gca acc att aat ctg g aaa tag caa aaa atg aca caa ggc	94 °C/30 s; 59 °C/30 s; 72 °C/30 s, 34 cycles
SRY-8299 (SRY-4064)	509 pb: *Bsr*BI 362 bp/147 bp	aca gca cat tag ctg gta tga c tct ctt tat ggc aag act tac g	94 °C/30 s; 62 °C /30 s; 72 °C/60 s, 33 cycles
SRY-2627 (SRY-2628)	1242 pb: *Hgi*AI (*Bsi*HKA 1) 298 bp/944 bp	agg tct ttt ttg cct tct ta atg cac ggt ttc ttt tga	94 °C/30 s; 52 °C /30 s; 72 °C/120 s, 33 cycles
M9	341 pb: *Hin*fI 182 + 93 + 66 pb (C)/ 248 + 93 pb (G)	aaa acc taa ctt tgc tca agc gca gca tat aaa act ttc agg	94 °C/30 s; 58 °C /30 s; 72 °C/30 s, 33 cycles
DYS257	394 pb: *Ban*I 182 + 106 + 63 + 43 pb (G)/ 288 + 182 + 106 + 63 + 43 pb (A)	gaa ctt gtc ggg agg caa t tga tac act tcc tcc ttt agt gg	94 °C/30 s; 60 °C /30 s; 72 °C/30 s, 33 cycles

Inuit Eskimos, (39) Irish, (40) Italian, (41) Japanese, (42) Kazakhs, (43) Khoisan, (44) Korean, (45) Latvian, (46) Lithuanian, (47) Manchurian Evenk, (48) Mari, (49) Melanesian, (50) Mixtecs, (51) Mongolian, (52) Navajo, (53) Ngöbe, (54) North African, (55) Northern Portuguese, (56) Northern Swedish, (57) Norwegian, (58) Oroqens, (59) Ossetian, (60) Papua New Guinean, (61) Pima, (62) Polish, (63) Pueblos, (64) Pygmies,

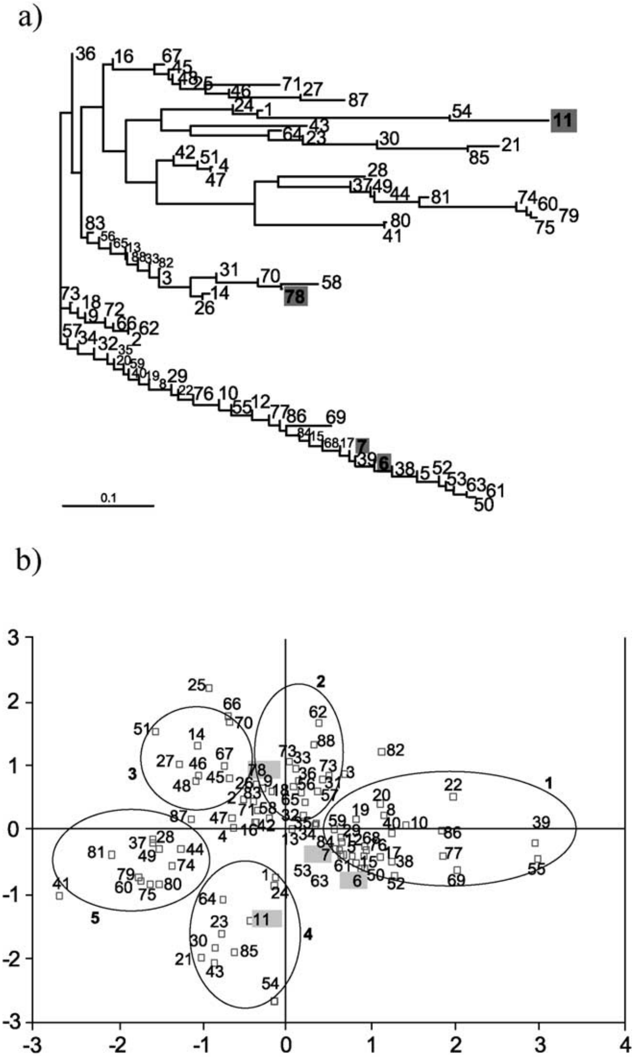

Fig. 1. (a)Neighbour-joining tree illustrating human phylogenetic relationships between Basque, Georgian and Berber populations with other worldwide populations. (b) Human phylogenetic relationships between Basque, Georgian and Berber populations with other worldwide populations in a multidimensional scaling analysis. (1) Southwest European populations, (2) North European populations, (3) East European populations, (4) African populations, (5) Asian populations.

(65) Romanian, (66) Russian, (67) Saami, (68) Scottish, (69) Selkup, (70) Siberian Evenks, (71) Siberian Skimos, (72) Slovakian, (73) Slovenian, (74) South Chinese, (75) Southeast Asian, (76) Southern Portuguese, (77) Spanish, (78) Swanetia, (79) Taiwan, (80) Tibetan, (81) Tundra Nentsi, (82) Turkish, (83) Ukranian, (84) Wayu, (85) West Bantus, (86) West Scottish, (87) Yakuts, (88) Yugoslavian (Fig. 1).

3. Results

The haplotype frequencies found in the samples of the studied populations are shown in Table 2. The highest frequencies in Basque, Georgian and Berber populations correspond to different haplotypes. In this way, the Basque populations have the highest frequency of haplotype V that has not been detected in Georgia populations. The Berber population sample exhibits a unique haplotype that does not appear in Georgia and it is rarely found in the Basque Country. Such dissimilar results are indicating great differences among these populations.

Of the 10 haplotypes determined by the eight analysed markers, 8 have been found in the Basque, Georgia and Berber populations (Table 2).

The phylogenetic analysis was performed by using all analysed markers, except for SRY2627. Their results confirm the observed differences. In the neighbor-joining tree obtained from genetic distances of Reynolds (Fig. 1a), the Basque autochthonous populations are on the occidental European Caucasoid branch, while Swanetia (Georgia) is in the east European branch. The Berber sample is on the North African populations branch. The differences between these groups are also observed using the χ^2 distance. The multidimensional scaling analysis (Fig. 1b) shows five principal groups. Three groups correspond to southwest, north and east European populations. The Basque populations are into the southwest European population group and the Georgian populations into the east European group. A fourth group is the African populations. The Berber population is placed into the fourth group. Asian populations are the fifth group, but none of the here studied populations belongs to this group.

In summary, our results do not show genetic similarities in the paternal lineages between Basques and Georgians, nor between Basques and Berbers.

Table 2
Haplotype frequencies of SNPs and YAP here studied in Basque, Georgian and Berber populations

Haplotype	SRY 1532	YAP	M9	SRY 8299	Tat	DYS 257	SRY 2627	Sy81	Basques Arratia	Basques Goierri	Residents Basque Country	Residents Georgia	Swanetia Georgia	Berbers
II	1	0	0	0	0	0	0	0	0.03	0.12	0.22	0.81	0.92	–
IV	1	0	1	0	0	0	0	0	–	–	0.05	–	–	–
3G	1	1	0	0	0	0	0	0	0.03	–	–	–	–	–
1D	0	0	1	0	0	1	0	0	–	–	–	0.19	0.08	–
III	1	1	0	1	0	0	0	0	0.09	0.04	0.05	–	–	1.00
V	1	0	1	0	0	1	0	0	0.79	0.72	0.60	–	–	–
5	1	1	0	1	0	0	0	1	–	–	0.03	–	–	–
22	1	0	1	0	0	1	1	0	0.06	0.12	0.05	–	–	–

4. Discussion and conclusions

Previous studies of chromosome Y have observed no relationships between Basque and Georgian populations [7]. The results of nuclear DNA studies in Basques and Berbers are contradictory [2,3]. On the other hand, some linguistic studies find certain similitudes between the Basque language and Georgian and Berber languages [1,4,6].

Our results do not confirm the relationships among Basque, Georgian and Berbers as they have been suggested by linguistic studies, or among Basques and Berbers as the studies of nuclear DNA have pointed out, but they are coincident with the genetic results observed in Georgian population by Rosser et al. [5] and Bosch et al. [3].

In conclusion, the study of the paternal lineages by means of the haplotypes of seven SNPs of the chromosome Y and the marker YAP does not show closed phylogenetic relationship among the Basque, Georgian and Berber populations.

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