International Congress Series 1288 (2006) 168–170

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Y-chromosome genetic structure in a sub-Apennine population of the Marches (central Italy): Analysis by SNP and STR polymorphisms

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Abstract. In order to define the Y-chromosome genetic structure in Apennine populations, 17 Y-chromosome microsatellites and 37 Y-single nucleotide polymorphisms were typed in 81 subjects living in Fabriano and Urbino, two small towns in the upland area of the Marches (central Italy), speaking different dialects and submitted to a limited genetic flow. © 2005 Published by Elsevier B.V.

Keywords: Y-chromosome; Short tandem repeat; Single nucleotide polymorphism; Haplotype; Haplogroup; Italy; The Marches

1. Introduction

In this study Y chromosomal markers were examined in 81 healthy male donors, unrelated and with different surnames, selected from two hinterland areas of the Marches region, Fabriano ($n=44$) and Urbino ($n=37$). All samples were genotyped for 17 Y-STRs using AmpFlSTR® Yfiler™ (AB) that allow co-amplification of the core set of European Minimal Haplotype, and other eight loci (DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and Y GATA H4).

37 Y-SNPs in six home-made multiplex PCRs were typed for all the 81 samples in order to obtain a highly discriminative picture of haplogroups spanning these micro-populations.

2. Materials and method

DNA was extracted from peripheral blood using phenol–chloroform protocol.

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0531-5131/ © 2005 Published by Elsevier B.V.
doi:10.1016/j.ics.2005.09.159

Y-STRs were amplified according to the manufacturer (AmpFlSTR® Yfiler™ PCR Amplification Kit, AB Applied Biosystems) modifying the final volume in 5 μl. Amplification of 37 Y-SNPs in six multiplex PCRs and subsequent single base extension were carried out according to Onofri et al. [1].

Typing was accomplished by ABI PRISM 310 Genetic Analyzer (AB Applied Biosystems). According to ISFG recommendations [2], GATA H4 locus name was replaced in GATA H4.1 and 10 repeats were added to alleles nomenclature of the kit.

Statistical analyses were performed using Arlequin software ver 2.000 [3]; DYS385 was excluded from calculation of haplotype diversity and AMOVA analysis.

3. Results and discussion

3.1. Y-STRs

In 81 subjects, 75 haplotypes were observed, 70 of which are unique. A duplication of the locus DYS458 was observed in one subject. The discrimination capacity, determined by dividing the number of observed haplotypes by the number of individuals, is 0.926 (minimum Ht = 0.826 only).

Haplotype diversity calculation resulted in a high level of differentiation; Urbino showed lower genetic variability (0.9940 ± 0.0086; pairwise differences = 9.8183 ± 4.5983) than Fabriano (0.9968 ± 0.0057; pairwise differences = 10.0497 ± 4.6821). Analysis of molecular variance (AMOVA) showed that variation among the two samples is 0.23% and 99.77% among individuals within the group, with a not statistically significant percentage of variation among the two areas, suggesting that there could be a background homogeneity.

3.2. Y-SNPs

A total number of 12 Y-SNP haplogroups were observed (Fig. 1). The most frequent one is R1b3, as previously observed in European populations [4].

Gene diversity is 0.7787 ± 0.0005 for Urbino and 0.8223 ± 0.0004 for Fabriano sampling groups.

Results of AMOVA showed that a statistically significant level of genetic differentiation exists among Fabriano and Urbino ($F_{ST} = 0.02523$, $P < 0.05$), which suggests a higher population differentiation if compared with other Italian population pairs [5].

A low Y-STR haplotypes variability within F*(x K, J2, I, H, G) and R1b3 Hgs in Urbino (0.9000 ± 0.1610 and 0.9714 ± 0.0389, respectively) and J2f and R1b3 Hgs in Fabriano (0.8333 ± 0.2224 and 0.9905 ± 0.0281) populations was observed, suggesting that a founder effect, even though weak, could have occurred in these confined mountain regions.

Haplogroups frequencies found in our sample were compared with those published for other Italian populations (Semino et al., north-central Italy sampling, and Di Giacomo et al., central-south

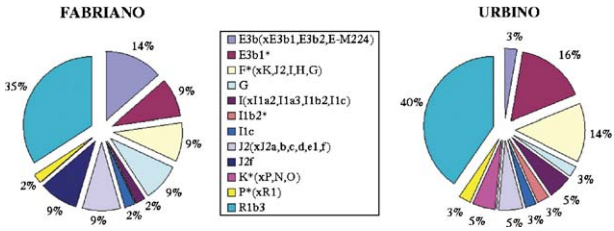

Fig. 1. Y-SNPs haplogroup frequencies observed in the two sampling groups.

Table 1
Haplogroup frequencies of Marche population compared to other heterogeneous Italian samplings

Haplogroup	Frequency		
	Marche ($n=81$)	Semino et al. ($n=50$)	Di Giacomo et al. ($n=524$)
R1b	0.370	0.620	0.364
DE	0.209	0.020	0.158
J2	0.123	0.140	0.206
I	0.074	0.080	0.065
G	0.062	0.100	0.063
R1a	–	0.040	0.034
Other	0.162	–	0.110

predominant, Table 1). This analysis showed that the Marches sample lies close to central-south Italian populations ($F_{ST}=0.0086$).

References

[1] V. Onofri, et al., Development of multiplex PCRs for evolutionary and forensic applications of 37 human Y chromosome SNPs, Forensic Sci. Int. (2005 May 13), doi:10.1016/j.forsciint.2005.03.014.
[2] L. Gusmao, et al., DNA Commission of the International Society for Forensic Genetics (ISFG): an update of the recommendations on the use of Y-STRs in forensic analysis, Forensic Sci. Int. (2005), doi:10.1016/j.forsciint.2005.04.002.
[3] S. Schneider, D. Roessli, L. Excoffier, A software for population genetics data analysis, Arlequin Version 2.000, University of Geneva, 2000.
[4] O. Semino, et al., The genetic legacy of Paleolithic *Homo sapiens sapiens* in extant: a Y chromosome perspective, Science 290 (2000) 1155–1159.
[5] F. Di Giacomo, et al., Clinal patterns of human Y chromosomal diversity in continental Italy and Greece are dominated by drift and founder effects, Mol. Phylogenet. Evol. 28 (2003) 387–395.