

The distribution of Y-chromosomal haplotypes and haplogroups in two population samples from the Romagna region (North Italy): Differences between urban (Rimini) and rural area (Valmarecchia)

G. Ferri*, S. Ceccardi, F. Lugaresi, F. Ingravallo,
C. Bini, A. Cicognani, S. Pelotti

*Department of Medicine and Public Health, Section of Legal Medicine,
University of Bologna, via Irnerio 49, 40 126 Bologna, Italy*

Abstract. We have studied the distribution of Y chromosomal haplotypes and haplogroups in two population samples from the Romagna region (North Italy) by analyzing male-specific markers that reflect past and recent history: SNPs and STRs. The population samples were collected in the urban area of Rimini, an ancient port in Roman age and in the near rural area of Valmarecchia, that is more isolated and geographically out of ancient trading ways. We analyzed 11 Y STRs by a commercial kit and 20 binary polymorphisms by minisequencing analysis. In spite of the different historical records and geographic locations, the two population samples showed high genetic affinities likely due to modern demographic migrations that might have eroded the past genetic substructure. © 2005 Elsevier B.V. All rights reserved.

Keywords: Population genetics; Y-chromosome; SNP; STR; Romagna area

1. Introduction

The simultaneous analysis of SNPs and STRs markers on the nonrecombinant region of the Y chromosome have been widely used to infer population history [1]. We have studied

* Corresponding author. Tel.: +39 051243250; fax: +39 051248896.

E-mail address: susi.pelotti@unibo.it (G. Ferri).

the distribution of Y chromosomal haplotypes and haplogroups in two different population samples from the Romagna region (North Italy) by SNPs and STRs analysis. One population sample was collected in the urban area of Rimini, an ancient port in Roman age and the other one in the near rural area of Valmarecchia, more isolated and geographically out of ancient trading ways. The aim of this study was to analyse the microgeographic heterogeneity of Y chromosome in a Northern Italian region and to link it to geographical and historical perspectives.

2. Materials and methods

163 unrelated individuals (98 from Rimini and 65 from Valmarecchia) were analyzed for 11 Y STRs (DYS391, DYS389I, DYS389II, DYS439, DYS438, DYS437, DYS19, DYS392, DYS393, DYS390 and DYS385) using the protocol recommended by the Power-Plex Y System (Promega, Madison, WI) and for 20 binary polymorphisms, grouped in three multiplexes for determining the most frequent haplogroups, by minisequencing analysis using published primer sequences [2]. PCR multiplexes were performed in a 25- μ l volume as reported by Brion et al. [2]. Multiplex SNaPshot reaction were performed in 8 μ l volume, including 2 μ l of multiplex SNaPshot kit (Applied Biosystem) and 1.5 μ l of cleaned PCR product, as by manufacturer. Before ABI 310 analysis, snapshot products were incubated with 1 U of SAP (Amersham Biosciences). Microsatellites haplotype and binary marker haplogroup frequencies were calculated and Arlequin 2.0 package [3] was used to estimate statistical parameters like analysis of molecular variance (AMOVA) and exact test of population differentiation.

3. Results and discussion

A total of 149 different haplotypes were found among 163 individuals and 138 haplotypes were observed in just a single male reflecting high haplotype STRs diversities. 11 haplotypes were non-unique (one belonging to E3b1 haplogroup observed in four individuals, one belonging to E3b1 haplogroup observed in three individuals, nine belonging to R1b, E3b1 and G haplogroups observed in two individuals) and 6 haplotypes, belonging to E3b1, R1b and G

Table 1
Haplogroup frequencies in Rimini and Val Marecchia samples

Rimini		Val Marecchia	
Haplogroup	Frequencies	Haplogroup	Frequencies
E3b1	11.22%	E3b1	18.46%
E3b3a	4.08%	E3b*	1.54%
G	7.14%	G	10.77%
I*	4.08%	I*	3.08%
K*	1.02%	I1b2	1.54%
J2	17.35%	J2	15.38%
N3	2.04%	R1a	1.54%
R1a	1.02%	R1b	46.15%
R1b	51.02%	K2	1.54%
R1*	1.02%		

Table 2

Gene diversity value of the R1b and E3b haplogroups

R1b	80 haplotypes	75 original
Gene Diversity	0.9984 ± 0.0023	
E3b	24 haplotypes	16 original
Gene Diversity	0.9565 ± 0.0250	

haplogroups, were shared between Rimini and Valmarecchia samples. Rimini sample showed 92 different haplotypes and 87 are unique. Val Marecchia on 63 different haplotypes showed 61 unique haplotypes. Table 1 shows haplogroups distribution. By the analysis of molecular variance (AMOVA), performed considering separately STRs and SNPs variations, no significant differences between the two population samples were observed and ~99–100% of the variance remained within the population. Haplogroup R1b was found to be the most dominant Y lineage in all samples. E3b3a (M34) haplogroup was found exclusively in Rimini population. Gene diversity calculated inside R1b and E3b haplogroups showed a reduced heterogeneity in E3b, suggesting its relatively recent colonization (Table 2).

Curiously in the Rimini population we found two unrelated individuals typed for rare cluster β of E3b1 haplogroup with 10-repeat allele at DYS439. This finding is not easily explained because of the cluster β of M78 mutation is common in Northwestern Africa and outside this region was observed only in five European subjects [4].

In spite of the different historical records and geographic locations, the two population samples showed high genetic affinities likely due to modern demographic migrations that might have eroded the past genetic substructure.

Up to date this study represents the first report on Y distribution in Romagna area, nevertheless our estimate suggested that more molecular data, linked to surnames analysis, were necessary to further elucidate the genetic background of this area. An increasing sampling is in progress from the near valleys between Romagna and Tuscany, far from the sea and defined by the Appenino Mountain.

References

- [1] P. de Knijff, Messages through bottlenecks: on the combined use of slow and fast evolving polymorphic markers on the human Y chromosome, *Am. J. Hum. Genet.* 67 (2000) 1055–1061.
- [2] M. Brion, et al., Hierarchical analysis of 30 Y-chromosome SNPs in European populations, *Int. J. Leg. Med.* 119 (2005) 10–15.
- [3] S. Schneider, D. Roessli, L. Excoffier, Arlequin Ver. 2.000: A Software for Population Genetics Data Analysis, Genetics and Biometry Laboratory, University of Geneva, Switzerland, 2000.
- [4] F. Cruciani, et al., Phylogeographic analysis of haplogroup E3b (E-M215) y chromosomes reveals multiple migratory events within and out of Africa, *Am. J. Hum. Genet.* 74 (2004) 1014–1022.