

Research article

Microgeographic variation of Y-chromosome haplotypes in Italy

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GeFI's group of Y-chromosome characterization

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Abstract

Within an Italian collaborative exercise on the extended haplotype of the Y-chromosome, 1288 subjects were typed by the AmpFISTR Yfiler Amplification Kit (AB Applied Biosystems) and other 526 were typed by the PowerPlex Y[®] System (Promega). The sampling scheme included either a "regional" or a "local" recruitment, the first referring to individuals born in the region of the participating lab, the second referring to individuals coming from small villages. Total sample sizes were $N = 954$ and 860 , respectively. A significant decrease of haplotype diversity was found in the local samples. The results may be of interest in forensic applications of the Y-chromosome.

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1. Introduction

Up to now the haplotype diversity using nine Y-STRs comprising the so-called minimal haplotype loci was studied among worldwide population samples showing that there are significant portions of haplotypes in several populations which cannot be resolved. Evaluation of haplotype discrimination

capacity of 35 Y-STRs was recently evaluated and complete resolution of the pooled population was achieved by additional genotyping of further loci [1]. Y-STRs generating haplotypes were studied in 2001 by GeFI collaborative exercise on 1176 Italian individuals from different regions [2]. The typed loci were DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385 and a low degree of variations was shown among regions. In forensic genetics laboratories Y-STRs multiplex kit, based on 17 markers validated for forensic applications, have become widely used in the last years for the high power of discrimination at minimal samples consumption.

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Nine-locus haplotype	Northern Italy					Central Italy							Total counts	Expected counts	Freq.	Rank in Italy		
	Bergamo	Brescia	Cuneo	Rimini	Valmarecchia	Urbino	AscoliPiceno	Fabriano	Matelica	Buti	Col	Msb					Nor	Sezze
14-13-29-24-11-13-13-11-14	6	4	3		3	1				2		1	4	1	25	15.6	0.040	1
14-13-29-24-10-13-13-11-14	4	4	2			2				1					13	6.2	0.021	2
14-13-29-24-11-13-12-11-14			3							1			3	1	8	5.2	0.013	16
14-13-29-23-11-13-13-11-14	3	1	1			1						1			7	4.5	0.011	3
14-13-30-24-11-13-13-11-14	3	1	2							1					7	3.8	0.011	71
14-13-29-24-10-13-13-11-15	1	1									4				6	3.8	0.010	6
15-12-29-22-10-11-14-14-14	1	2	1	1								1			6	3.6	0.010	41
13-13-30-24-10-11-13-16-18				3	2										5	3.4	0.008	5
14-13-29-24-11-13-13-11-15	1	1	1	1						1					5	3.2	0.008	4
14-13-29-24-11-14-13-11-14		2	1	1						2					5	3.1	0.008	19
15-13-29-24-11-13-13-11-14	1	1		2							1				5	3.1	0.008	22
13-13-30-23-10-11-13-16-18			1	2									1		4	2.8	0.006	48
14-13-29-24-11-13-14-11-14	1	2					1								4	2.8	0.006	65
14-13-29-25-11-13-13-11-14	1		2					1							4	2.8	0.006	7
15-12-28-23-10-11-12-13-17			1									3			4	2.8	0.006	#N/A
13-12-29-24-10-11-13-17-18			2				1								3	2.7	0.005	128
13-13-29-24-11-13-13-11-14		2									1				3	2.4	0.005	168
13-13-30-23-10-11-13-16-17			1	1	1										3	2.2	0.005	#N/A
13-13-30-24-09-11-15-15-17								3							3	2.0	0.005	#N/A
13-13-30-24-10-12-13-16-18	1		1			1									3	2.0	0.005	#N/A
14-12-28-23-11-11-12-13-17												3			3	2.0	0.005	#N/A
14-12-29-24-10-11-13-17-18	2	1													3	2.0	0.005	#N/A
14-13-28-24-10-13-13-11-14							2	1							3	2.0	0.005	27
14-13-28-24-11-13-13-11-14	1	1										1			3	2.0	0.005	344
14-13-29-23-10-13-13-11-14				1	2										3	2.0	0.005	9
14-13-29-23-11-13-13-11-15			2	1											3	2.0	0.005	13
14-13-30-25-11-13-13-11-14				3											3	2.0	0.005	#N/A
14-14-31-24-11-13-13-11-15					3										3	2.0	0.005	#N/A
15-12-28-24-10-11-12-14-17		1	1								1				3	2.0	0.005	83
Others	20	63	66	69	50	29	34	36	9	28	16	26	19	16	481	448	0.762	
Total	22	87	86	98	65	40	38	44	9	38	21	29	35	19	631		1	

Fig. 1. The most frequent “minimal haplotypes” in 14 local samples from central and northern Italy. #N/A: not present in the Italian database.

This further GEFI collaborative project was designed for studying the diversity of 17-locus Y-STR profiles, usually used in casework, on different Italian population groups, sampling by regional or local ways for a total of 1288 typed samples to determine individual loci gene diversity, multiplex discriminatory capacity and to increase data for reference database. In addition a total of 526 samples were typed for 12 loci by a few number of laboratories and the results were collected to increase the minimal haplotype Italian database.

2. Materials and methods

Participating laboratories were asked to type at least 100 unrelated individuals born in their region for 17 loci by Y filer kit. Blind control samples were prepared for each laboratory. Laboratories were left free to use their preferred DNA extraction methods. PCR and analysis of amplified products were performed according to the manufacturer’s recommendations. In addition other 526 Italian samples were typed by the PowerPlex Y[®] System (Promega).

3. Results and discussion

Within an Italian collaborative exercise on the extended haplotype of the Y-chromosome, 1288 subjects were typed by the AmpFISTR YFiler Amplification Kit (AB Applied Biosystems) and other 526 were typed by the PowerPlex Y[®] System (Promega). The sampling scheme included either a “regional” or a “local” recruitment, the first referring to individuals born in the region of the participating lab, the second referring to individuals coming from small villages. In the second case, only non-isonymous subjects were sampled. Fig. 1 shows the 9-locus haplotype counts in 631 individuals from 14 local samples compared with the expected counts in a sample of the same size if

Seventeen-locus haplotype	Bergamo	Brescia	Cuneo	Urbino	AscoliPiceno	Fabriano	Matelica	Buti	Col	Msb	Nor	Sezze	Total
14-13-29-24-10-13-13-11-15-14-13-12-18-15-18-23-12													4
14-14-31-24-11-13-13-11-15-14-12-12-19-16-18-27-12				3									3
13-13-29-24-11-13-13-11-14-15-12-11-19-15-16-23-12			2										2
13-13-30-25-10-11-13-17-17-14-10-11-20-15-15-24-12										2			2
13-13-30-25-11-11-13-17-17-14-10-13-20-15-17-22-12						1			1				2
13-13-32-22-11-11-13-17-17-14-10-12-20-15-14-21-13						2							2
14-12-28-23-10-11-13-13-16-10-10-20-14-15-23-10			2										2
14-12-28-23-10-11-13-13-14-16-10-12-20-14-15-21-11			2										2
14-12-28-23-11-11-12-13-17-14-09-12-21-14-19-21-11											2		2
14-13-28-24-10-13-13-11-14-15-11-11-19-17-16-23-12							2						2
14-13-29-24-11-13-12-11-14-15-12-13-19-15-15-23-12												2	2
14-13-29-24-11-13-13-10-13-15-12-12-19-15-18-23-12												2	2
14-13-29-24-11-13-13-11-14-15-12-14-20-16-17-23-12												2	2
14-13-29-24-11-13-13-11-14-15-13-13-19-16-18-23-11												2	2
14-13-30-23-10-11-12-13-15-15-09-12-20-17-17-21-11						2							2
15-12-28-23-10-11-12-13-17-16-09-12-19-13-17-23-11											2		2
15-12-29-22-10-11-14-13-17-15-11-10-21-15-16-20-12				2									2
15-14-30-23-10-11-12-12-16-15-09-11-20-15-16-23-11											2		2
Unique haplotypes	22	78	79	35	38	37	9	38	16	27	21	19	419
Total	22	78	85	40	38	44	9	38	21	29	35	19	458

Fig. 2. Non-unique 17-locus haplotypes in 12 local samples from northern and central Italy.

it were randomly sampled from the general Italian population. The expected counts were obtained by numerical resampling of the Italian database. For example, the first two most frequent haplotypes among the local samples display the highest rank in Italy also; however, their relative frequency is higher in the local samples (observed counts 38 vs. expected counts 21.8). In general, the haplotype frequency distribution is biased in the local samples towards a lower number of haplotypes with higher frequency. Fig. 2 shows the non-unique 17-locus haplotype counts in 12 local samples (two local samples were not typed with the 17-locus kit). It is remarkable that all non-unique haplotypes but one are present in a single local sample.

Conflict of interest

None.

References

- [1] H. Roding, L. Roewer, A. Gross, T. Richter, P. de Knijff, M. Kaiser, W. Brabetz, Evaluation of haplotype discrimination capacity of 35 Y-chromosomal short tandem repeat loci, *Forensic Sci. Int.* 174 (2008) 182–188.
- [2] S. Presciuttini, A. Caglià, M. Alù, A. Asmundo, L. Buscemi, L. Caenazzo, E. Carnevali, E. Carra, Z. De Battisti, F. De Stefano, R. Domenici, A. Piccinini, N. Resta, U. Ricci, V.L. Pascali, Y-chromosome haplotypes in Italy: the GEFI collaborative database, *Forensic Sci. Int.* 122 (2001) 184–188.