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Announcement of population data

Frequency assessment of 25 SNPs in five different populations

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ABSTRACT

Allele and genotype frequencies of 25 SNPs previously selected and validated for forensic purposes were assessed in 250 unrelated individuals originating from five different countries of Europe (Spain, Croatia, Bulgaria, Turkey and Serbia). All the SNPs generated extremely low F_{st} values confirming our previous results on Italian, African (Benin) and Asian (Mongolian) populations. As a consequence of such F_{st} values we observed similar values of random match probability across the populations: 2.26×10^{-10} in the Spanish population, 2.13×10^{-10} in the Croatian population, 4.21×10^{-10} in the Bulgarian population, 2.52×10^{-10} in the Serbian population and 1.46×10^{-10} in the Turkish population.

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1. Population

Two hundred and fifty unrelated individuals from five different populations: Spanish ($n = 50$), Croatian ($n = 50$), Bulgarian ($n = 50$), Serbian ($n = 50$), and Turkish ($n = 50$).

2. Extraction

Genomic DNA was extracted from whole blood by the QIAamp DNA Blood Mini Kit (QIAGEN Inc., Valencia, CA).

3. SNPs

A number of 25 SNPs were previously selected on the basis of a number of selection criteria reported and validated elsewhere [1,2]. The 25 SNPs are: rs1779866, rs1922807, rs2278741, rs905213, rs11242909, rs3130315, rs1075665, rs7740233, rs10866988, rs585070, rs1506981, rs1533800, rs1981752, rs478347, rs9562080, rs911621, rs999842, rs8033863, rs886528, rs154659, rs2317225, rs873289, rs11881170, rs380011 and rs2267628.

4. SNP typing

DNA typing was performed with 10 ng of target DNA using Pre-design TaqMan[®] Genotyping assays (Applied Biosystems, Foster

City, CA). Fluorescence was detected using an ABI 7500 Sequence Detection System and genotypes were manually scored using Sequence Detection Software 2.0 (Applied Biosystems).

The list of the SNPs together with their relative chromosome localizations, assay ID and context sequence (including regions where probes FAM and VIC have been designed) are reported in Table 1.

5. Genotyping confirmation and statistical analysis

Allele frequencies were calculated by direct counting. Genotypes of 50 random samples were confirmed by direct sequencing. All statistical and forensic parameters were calculated [3] using DNAVIEW[™] 27.19. Statistical independence of selected markers was assessed by calculating linkage disequilibrium (LD) as r^2 [4] using LD plotter software (available at <https://www.pharmgat.org/Tools/pbtoldplotform>). Divergence from Hardy–Weinberg equilibrium (HWE) was examined using the on-line calculator (<http://ihg2.helmholtz-muenchen.de/cgi-bin/hw/hwa1.pl>). Inter-population genetic distance was assessed by calculating F_{st} for each marker (http://genepop.curtin.edu.au/genepop_op6.html).

6. Results

No departure from the Hardy–Weinberg equilibrium was detected. To exclude the dependence of markers from each other we calculated the pairwise LD values reported as r^2 . All selected SNPs showed r^2 values near to zero (complete independence).

The frequencies of the minor allele (MAF) of the SNPs are reported in Table 2.

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Table 1

Name, chromosomal localization, nucleotide position, assays ID and context sequence including FAM/VIC probes of all SNPs selected.

SNPs	chr	Nucleotide position	Assay ID	Context sequence including FAM/VIC probe
rs1779866	1	61254987	C_2718124_20	TGCTTTGGACCCAGCTGCATAATC [C/G] AAGATCCTCTCCTTAATCCATT
rs1922807	2	77516772	C_11940372_10	ATATTAAGCGGTACCAAAATTA [C/T] TGAATCATCTATTGATAAAACATGC
rs2278741	2	121465537	C_3254038_20	GAATGTAGCCCTTCTGAAAGAA [C/G] ACTTGTTCCTAAATACCTCGGGCT
rs905213	5	91995945	C_1454141_10	TAACGTAACCCGAAGTCTTTAG [G/A] ACTTATTAATGATTTCGCCAC
rs11242909	6	422065	C_1354181_10	TTGTGTGATATCAA CTTAGACCA [G/A] TAGCTTCTTTTCATGAATTTT
rs3130315	6	32328663	C_3293919_10	CTGCAAAGATATGGGTTTGGTT [A/G] GAGAGTGAGATGCTGATGTGTAAGG
rs1075665	6	88423321	C_11568848_10	TTGGATTTACCCCTATTGATGCT [A/G]CTGGTTTTGTATTAAGCAGAAAC
rs7740233	6	150915459	C_1201558_10	GCCCTTCTGCTTGTTTTAGGAC [C/T] TGAGACAAAGGGAGTTTTATAAGAA
rs10866988	8	5235745	C_30890631_10	TGCTTTGATGTGTGCTTCTCC [C/G] AAGGCCAAAACGGCCAAAAGAGT
rs585070	10	113617876	C_794076_10	CTAAATTTGTGTTAAGCTTCCCTA [C/T] GTATAAAACATTCCTAAGTTTGT
rs1506981	11	6735647	C_9599956_10	TATGGAGGCTATAAGAGCCACC [A/G] GTTTCAATTCAGTATCTAAATCAC
rs1533800	11	33781074	C_12032648_10	AGGTCAGCAGGAACCTCAGACTAGA [G/T] CTGGGATCTGGAATCCCGCCACA
rs1981752	12	16430035	C_12044306_10	GCAAGTTGGAGGGCTGCTTTAA [C/T] ACATACAGTTCTTTTGGAGTTTGT
rs478347	12	128956977	C_673674_20	ATTCAAGTACTGCAGGGGCTATGC [A/C] ACTGGTGGATGAGAGGACACTTCA
rs9562080	13	113941879	C_32266871_10	ATCTCTGAGCATCTCTGGCTGGG [C/G] CCTCACCTGCCCCACCGCCACA
rs911621	14	54195466	C_7593433_10	CTCTGAGATTCAGTTGCACACCTG [C/T] AAATCAAGAATGCTGGCTCACAGG
rs999842	15	20551713	C_9401442_10	GTGGATGAAAATCTCGCACCATCT [G/A] TGGACTTTTCAAAGCATACTTT
rs8033863	15	99857192	C_2030634_10	GGCTAATAACAATGCTGGTGGCAG [C/G] GCTGTGCCAGCAGTGCAGTATTTT
rs886528	16	3751557	C_8928292_10	CATGCTCAGGGCTCATTCCATCA [A/G] AGCCAGGATACAGACTCTGGACCA
rs154659	16	88194838	C_3224527_10	CCTGCAATTAGAGCTGACCTTGC [C/T] GAGAATAGCCTGGTATGACTGCC
rs2317225	17	2593098	C_2593470_10	CAAGCCAAAACATCCTTCTGTGT [A/G] CCATTGCGTAGGCTCTGACTGTC
rs873289	19	1126396	C_9059193_10	GCCCCATTCACCCCTGAAATC [A/G] GAGTTGATTTCTCCATGACTTTGT
rs11881170	19	62864113	C_30857838_10	CAGATCAGGATAAAATTGAGCCTC [A/C] TTACTAGTTACTACGATTTTGAAG
rs380011	21	14542669	C_2401290_10	TTGAGTTCAACTGGGTACAAATC [C/T] TATTCTACCTGGAATCAGAGGCT
rs2267628	X	137559012	C_15872694_10	ACCAAGTCATTATAAGTTGCATCAT [C/T] TATTATGAATCACATCAGCAATTG

The genetic distance F_{st} coefficients among the 5 populations ranged between 0.0001 and 0.0608, while the mean value observed for all SNPs was 0.0187 ± 0.05 (Table 2).

The random match probability for each population were: 2.26×10^{-10} in the Spanish population, 2.13×10^{-10} in the Croatian population, 4.21×10^{-10} in the Bulgarian population, 2.52×10^{-10} in the Serbian population and 1.46×10^{-10} in the Turkish population.

7. Other remarks

Genetic data resulting from the typing of 250 unrelated individuals originating from 5 different European populations confirmed the high information content of the selected SNPs. The

mean heterozygosity values calculated for the panel of 25 SNPs in the five populations considered in this study are 0.45 for Spanish population, 0.44 for Croatian, Bulgarian and Turkish populations and 0.42 for Serbian population. These values are quite similar to those reported in a previous work [2] for Italian (0.46), Benin Gulf (0.48) and Mongolian populations (0.48). Finally the random match probability of a panel of 25 SNPs for each population analyzed in this work is quite similar to the RMP obtained for Italian population (4.96×10^{-10}), Benin Gulf population (5.30×10^{-11}) and for the Mongolian population (3.31×10^{-11}).

This paper has strictly followed the requirements of FSI: Genetics guidelines and the ISFG recommendations [5-7].

Access to the data: Data is available on <http://www.nacbo.net>.

Table 2Minor allele frequencies (MAF), mean heterozygosity and F_{st} values of all selected SNPs in Spanish, Croatian, Serbian, Bulgarian and Turkish populations.

SNPs	Spanish MAF	Croatian MAF	Serbian MAF	Bulgarian MAF	Turkish MAF	Mean marker Het (5 populations)	F_{st}
rs1779866	0.40 [C]	0.37 [C]	0.45 [C]	0.40 [C]	0.44 [C]	0.45	0.0084
rs1922807	0.45 [G]	0.49 [G]	0.44 [G]	0.41 [A]	0.40 [A]	0.40	0.0066
rs2278741	0.24 [G]	0.32 [G]	0.38 [G]	0.28 [G]	0.46 [G]	0.38	0.0085
rs905213	0.46 [C]	0.46 [C]	0.41 [C]	0.49 [C]	0.47 [T]	0.55	0.0043
rs11242909	0.26 [C]	0.32 [C]	0.21 [C]	0.27 [C]	0.29 [C]	0.33	0.0053
rs3130315	0.48 [A]	0.34 [G]	0.39 [G]	0.41 [G]	0.36 [G]	0.51	0.0133
rs1075665	0.31 [C]	0.36 [C]	0.27 [C]	0.30 [C]	0.37 [C]	0.45	0.0041
rs7740233	0.50	0.33 [C]	0.33 [C]	0.41 [C]	0.49 [C]	0.55	0.0181
rs10866988	0.47 [G]	0.48 [C]	0.46 [G]	0.48 [G]	0.49 [G]	0.44	0.0116
rs585070	0.49 [G]	0.50	0.47 [G]	0.40 [G]	0.44 [G]	0.43	0.0074
rs1506981	0.44 [G]	0.40 [G]	0.35 [G]	0.45 [G]	0.39 [A]	0.55	0.0255
rs1533800	0.16 [A]	0.33 [A]	0.39 [A]	0.16 [A]	0.32 [A]	0.29	0.0469
rs1981752	0.33 [C]	0.34 [C]	0.37 [C]	0.16 [C]	0.44 [C]	0.38	0.0358
rs478347	0.47 [G]	0.48 [G]	0.44 [G]	0.46 [T]	0.40 [T]	0.51	0.0031
rs9562080	0.43 [C]	0.35 [C]	0.40 [C]	0.43 [C]	0.44 [C]	0.44	0.0069
rs911621	0.49 [C]	0.49 [T]	0.37 [T]	0.47 [T]	0.42 [T]	0.44	0.0006
rs999842	0.43 [C]	0.49 [T]	0.41 [C]	0.43 [T]	0.49 [C]	0.48	0.0052
rs8033863	0.35 [C]	0.42 [C]	0.31 [C]	0.38 [C]	0.37 [C]	0.46	0.0010
rs886528	0.49 [C]	0.30 [C]	0.41 [C]	0.36 [C]	0.40 [C]	0.49	0.0101
rs154659	0.20 [C]	0.17 [C]	0.25 [C]	0.16 [C]	0.23 [C]	0.27	0.0045
rs2317225	0.42 [G]	0.40 [G]	0.38 [G]	0.48 [G]	0.43 [G]	0.47	0.0068
rs873289	0.42 [C]	0.48 [C]	0.41 [T]	0.46 [C]	0.45 [C]	0.47	0.0033
rs11881170	0.44 [C]	0.46 [C]	0.46 [A]	0.37 [C]	0.47 [A]	0.55	0.0072
rs380011	0.42 [C]	0.39 [C]	0.29 [C]	0.34 [C]	0.40 [C]	0.44	0.0008
rs2267628	0.49 [G]	0.38 [G]	0.45 [G]	0.46 [G]	0.40 [G]	0.26	0.0102

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