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INTRODUCTION

- As an improvement in efficiency and in Human Discrimination Power, the new six dye multiplex kit PowerPlex® Fusion 6C System, by Promega, available for human identification can co-amplify 27 loci, in a single reaction, have been introduced in the last years with great success [1].
- This kit allows the amplification and detection of autosomal loci included in the expanded Combined DNA Index System CODIS, plus the loci Penta D, PENTA E and SE33 as well as Amelogenin for gender determination. Furthermore, this kit includes three Y –STRs (DYS391, DYS576 and DYS570), allowing allelic attribution in a total of 27 loci.
- This genetic markers extension satisfies not only CODIS but also European Standard Set recommendations. Thinking about continuous human migration movements, especially in a very cosmopolitan region like Lisbon and south of Portugal, and also, in keeping population studies and actualized STR databases we decided to update our previous studies.

METHODS

- Our sample is composed of 605 unrelated individuals, from paternity testing with laboratory identity anonymised. DNA was extracted by Prep-n-go Buffer™(Thermo-Fisher Scientific). [2]
- PCR amplification was performed with PowerPlex® Fusion 6C System, according to manufacturer's guidelines. Fragment analysis was carried out in an Applied Biosystems® 3500 Genetic Analyser. Electrophoresis results were analysed with GeneMapper® ID-X v1.4.[3]
- Autosomic STRs allele frequencies and population statistics, including Hardy-Weinberg equilibrium p-values from exact test probabilities. Forensic Parameters were analysed with PowerStats v.2.1. [4]
- In conclusion, our population information was updated in order to apply most recent data in our casework weight of evidence.

DISCUSSION

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RESULTS

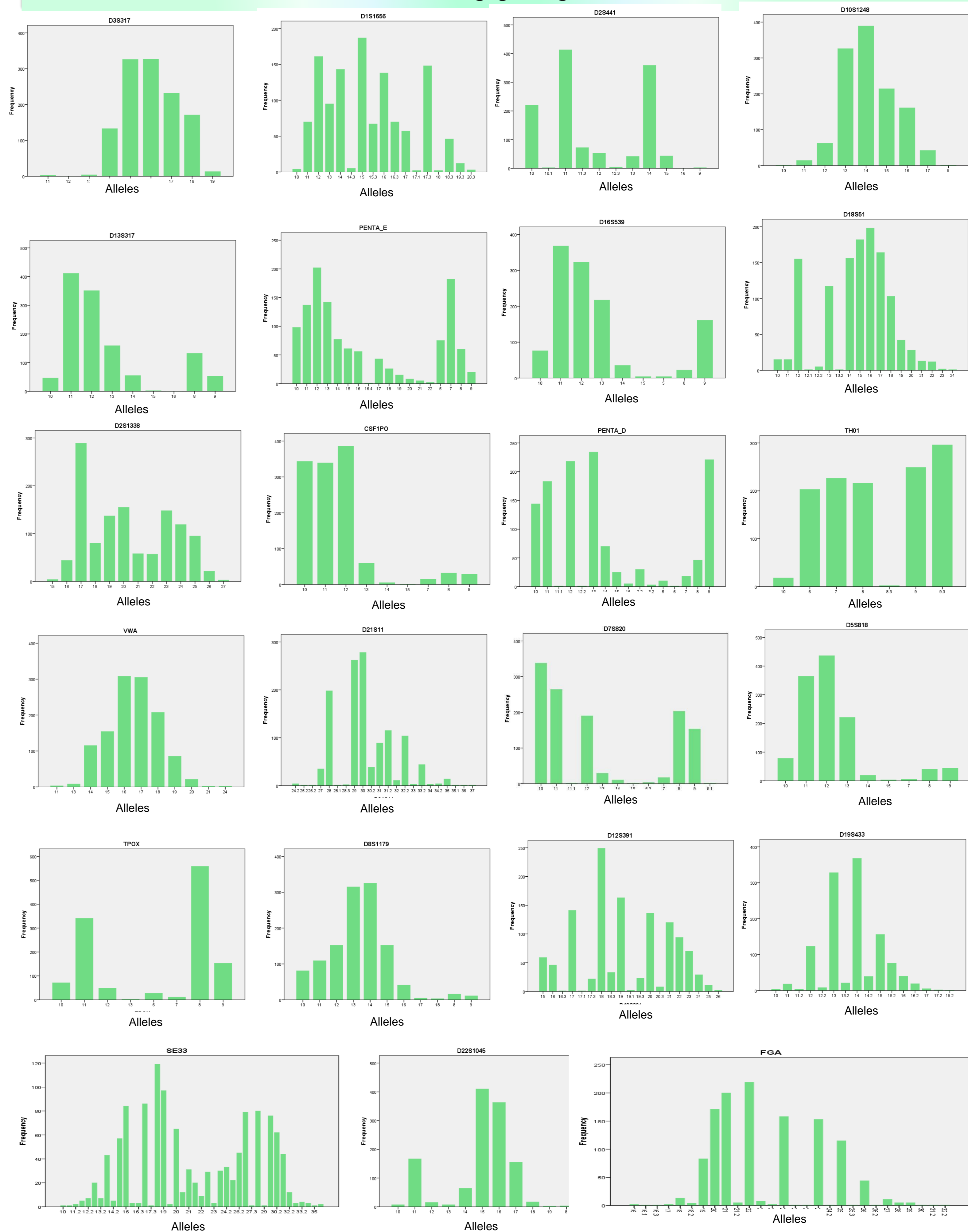


Fig.1-Graphical representation of allele frequencies on all autosomic markers included in the Powerplex® Fusion 6C (Promega)

Table 1- Forensic parameters and Hardy-Weinberg equilibrium p-values from exact test probabilities results obtained in all autosomic STRs included in Powerplex® Fusion 6C

	MP	PD	PIC	PE	TPI	HO	HEO	HEE	HW
D3S1358	0,0803	0,9200	0,7500	0,5890	2,4300	0,2060	0,7940	0,7861	0,2318
D1S1656	0,0470	0,9530	0,8200	0,6650	3,0300	0,1650	0,8350	0,8975	0,8605
D2S441	0,1270	0,8730	0,6800	0,4650	1,8000	0,2770	0,7230	0,7552	0,0167
D10S1248	0,0860	0,9140	0,7400	0,5170	2,0300	0,2460	0,7540	0,7718	0,3648
D13S317	0,0940	0,9060	0,7300	0,5550	2,2300	0,2240	0,7760	0,7665	0,3569
Penta E	0,0200	0,9800	0,8900	0,7980	5,0600	0,0990	0,9010	0,8999	0,8014
D16S539	0,0840	0,9160	0,7500	0,6010	2,5100	0,1990	0,8010	0,7819	0,0685
D18S51	0,0290	0,9710	0,8700	0,7150	3,5700	0,1400	0,8600	0,8811	0,1071
D2S1338	0,0300	0,9700	0,8600	0,6680	3,0500	0,1640	0,8360	0,8731	0,0036
CSF1PO	0,1240	0,8760	0,6900	0,5340	2,1200	0,2360	0,7640	0,7361	0,2232
Penta D	0,0450	0,9550	0,8300	0,6930	3,2200	0,1510	0,8490	0,8543	0,1097
TH01	0,1060	0,8940	0,7100	0,5580	2,2500	0,2230	0,7770	0,8034	0,8690
vWA	0,0630	0,9370	0,7900	0,6550	2,9300	0,1710	0,8290	0,8126	0,5979
D21S11	0,1000	0,9000	0,7200	0,4970	1,9400	0,2580	0,7420	0,8490	0,0893
D7S820	0,0690	0,9310	0,7700	0,6140	2,6000	0,1920	0,8080	0,8054	0,9146
D5S818	0,1060	0,8940	0,7000	0,4690	1,8200	0,2740	0,7260	0,7401	0,0649
TPOX	0,1430	0,8570	0,6400	0,3960	1,5600	0,3210	0,6790	0,6871	0,0067
D8S1179	0,0620	0,9380	0,7900	0,6100	2,5700	0,1940	0,8060	0,8152	0,1946
D12S391	0,0300	0,9700	0,8600	0,6680	3,0500	0,1640	0,8360	0,8888	0,5677
D19S433	0,1130	0,8870	0,6900	0,4770	1,8600	0,2690	0,7310	0,8008	0,0051
SE33	0,0320	0,9680	0,8600	0,6740	3,1200	0,1600	0,8400	0,9467	0,2567
D22S1045	0,1030	0,8970	0,7200	0,5460	2,1800	0,2290	0,7710	0,7571	0,2966
FGA	0,3300	0,6700	0,8500	0,7010	3,4000	0,1470	0,8530	0,8723	0,8534

MP-Matching Probability, PD-Discrimination Power, PIC-Polymorphic information content; PE-Exclusion Power, TPI-Typical Paternity Index, HO-Homozygosity, HEO-Observed Heterozygosity, HEE-Expected Heterozygosity, HW-Hardy-Weinberg p value.

- New Allele microvariants were detected in D1S1656 (17.1), D2S441 (10.1), Penta E (16.4), D18S51 (12.1, 12.2), Penta D (11.1, 12.2), D12S391 (17.1, 19.1) and FGA (16.1).
- Departure deviations from Hardy – Weinberg equilibrium were detected in D19S433 and TPOX, even after Bonferroni correction ($p < 0,05$)
- PENTA E (PD= 0,9800), D12S391 (PD=0,9700), D18S51 (PD=0,9710) and D2S1338 (PD=0,9700) are the most Polimorphic markers

References

- [1] Ensenberger MG, Lenz KA, Matthies LK, Hadinoto GM et al. Developmental validation of the PowerPlex® Fusion 6C System. *Forensic Sci Int Genet.* 2016 Mar;21:134-44.
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- [4] Excoffier, L. and H.E. L. Lischer (2010) Arlequin suite ver 3.5: A new series of programs to perform population genetics analyses under Linux and Windows. *Molecular Ecology Resources.* 10: 564-567.