

Announcement of Population Data

Allele frequencies of 14 STR loci in the population of Malta

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Abstract

Allele frequencies of 14 STR loci (D13S317, D16S539, D2S1338, vWA, TPOX, D18S51, D5S818, FGA, D8S1179, D21S11, D7S820, CSF1PO, TH01 and D3S1358) observed in the population of Malta are being reported. Polymerase chain reaction (PCR) amplification using the AmpFI STR[®] Identifiler kit was performed in a random sample of 157 subjects (314 chromosomes). Markers D2S1338, D18S51 and FGA had the highest power of discrimination (PD) values while TPOX was the least informative marker. Allele frequencies observed in the Maltese population were also compared with those of other populations from the Mediterranean region, Europe and Africa.

Our data is useful for anthropological and other comparative studies of populations and is powerful for forensic and paternity testing in the Maltese islands.

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Population: One hundred and fifty-seven unrelated individuals living in the island of Malta were randomly selected from paternity cases submitted to our facility. An informed consent was completed by each individual. Individuals that were not of a Maltese origin were excluded from the analysis. Buccal swabs or peripheral blood in EDTA containers were collected for DNA analysis.

DNA extraction: Genomic DNA was extracted from peripheral blood leucocytes by a modified salting out technique [1]. DNA extraction from buccal swabs was performed by using AccuPrep[™] Genomic DNA extraction kit (Bioneer Corporation, Daejeon, Korea).

PCR: Amplification of 14 STRs and Amelogenin was performed according to manufacturer's instructions using the AmpFI STR[®] Identifiler kit (Applied Biosystems, Foster City, CA) in a 12.5 µl total reaction volume.

Typing: Amplified products were analyzed with reference ladder using an ABI 3130 genetic analyzer (Applied Biosystems, Foster City, CA). Analysis of data obtained

from genetic analyzer was performed using GeneMapper[®] software v3.5.

Results: Refer to Table 1.

Quality control: Participation in proficiency testing by the College of American Pathologists (CAP) (<http://www.cap.org>) and forensic inter-laboratory testing organised by Collaborative Testing Services Inc. (<http://www.collaborativetesting.com/>).

Analysis of data: Forensic and paternity statistics including power of discrimination (PD) and power of exclusion (PE) were calculated with PowerStats v2.1 (Promega, Madison, WI, USA) [2]. Hardy–Weinberg equilibrium and heterozygosity were calculated for all loci using Linkage Utility Programs by J. Ott [3] (<http://linkage.rockefeller.edu/>). Deviation from Hardy–Weinberg equilibrium was also analysed using FSTAT (<http://www2.unil.ch/popgen/softwares/fstat.htm>) for those loci that deviated from Hardy–Weinberg.

Genetic relationships between populations coming from the Mediterranean region Europe and Africa were analysed using previously published STR data. Populations used were Sicilians [4], Italians [5], Greek [6], Spanish [7], Tunisian [8], Turkish [9], German [10], Slovenian [11] and Afri-

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Table 1
Allele frequencies of 14 STRs in the population of Malta ($n = 157$)

Allele	D13S317	D16S539	D2S1338	vWA	TPOX	D18S51	D5S818	FGA	D8S1179	D21S11	D7S820	CSF1PO	TH01	D3S1358
6					0.006								0.162	
7							0.003				0.016	0.003	0.162	
8	0.118	0.006			0.500		0.020		0.041		0.134	0.022	0.086	
9	0.105	0.121			0.103		0.046		0.010		0.080	0.061	0.242	
9.3													0.338	
10	0.070	0.035			0.110	0.013	0.030		0.054		0.271	0.274	0.010	
11	0.252	0.341			0.248	0.016	0.336		0.080		0.287	0.315		
12	0.318	0.341			0.032	0.178	0.352		0.115		0.169	0.252		
12.2		0.003												
13	0.096	0.115				0.151	0.197		0.347		0.035	0.045		0.003
14	0.038	0.038		0.109		0.171	0.010		0.175		0.010	0.029		0.054
15	0.003			0.141		0.125			0.118					0.269
16			0.038	0.215		0.086	0.003		0.054					0.199
17			0.218	0.282		0.115								0.295
18			0.128	0.186		0.076		0.010	0.003					0.170
19			0.099	0.048		0.030		0.076						0.003
19.2						0.003								
20			0.163	0.019		0.020		0.089						0.006
21			0.058			0.007		0.146						
22			0.058			0.010		0.219						
22.2								0.007						
23			0.135				0.003	0.203						
23.2								0.003						
24			0.067					0.106	0.003					
24.2										0.016				
25			0.035					0.099						
26								0.036						
27										0.042				
28										0.123				
29										0.252				
29.2										0.003				
30										0.248				
30.2								0.003		0.045				
31										0.071				
31.2										0.065				
32										0.016				
32.2										0.071				
33.2										0.045				
35										0.003				
Ho	0.797	0.740	0.870	0.808	0.667	0.875	0.723	0.855	0.810	0.842	0.793	0.758	0.770	0.771
HWp	0.547	0.188	0.585	0.436	0.367	0.538	0.999	0.387	0.980	<0.0001	0.503	0.664	0.599	0.767
MP	0.072	0.119	0.035	0.066	0.162	0.036	0.123	0.044	0.064	0.049	0.073	0.101	0.093	0.102
PD	0.928	0.881	0.965	0.934	0.838	0.964	0.877	0.956	0.936	0.951	0.927	0.899	0.907	0.898
PIC	0.77	0.70	0.85	0.78	0.62	0.86	0.67	0.84	0.79	0.82	0.76	0.71	0.73	0.73
PE	0.569	0.410	0.675	0.477	0.464	0.745	0.455	0.677	0.740	0.698	0.546	0.512	0.604	0.566
TPI	2.31	1.60	3.12	1.86	1.80	4.00	1.77	3.15	3.93	3.37	2.18	2.01	2.53	2.29

Ho, observed heterozygosity; HWp, Hardy–Weinberg equilibrium p -value; MP, matching probability; PD, power of discrimination; PIC, polymorphism information content; PE, power of exclusion; TPI, typical paternity index.

can (Equatorial Guinea) [12]. Genetic divergence was based on allele frequencies of five STRs (vWA, D18S51, D8S1179, D21S11, D3S1358) commonly tested in all populations. D_A genetic distance was calculated as described by Nei and colleagues [13], which is a more reliable estimate of evolutionary relationships between closely related populations than standard genetic distance (D_S) [14]. D_S and D_A were both used to construct phylogenetic trees by neighbour-joining (NJ) clustering [15]. Bootstrap re-sampling of 1000 replicates was performed to obtain the most reliable dendrograms. These analyses were performed using the software DISPAN [16].

Access to data: Complete data can be acquired upon request to chrisv@mlsbiodna.com and from website <http://www.mlsbiodna.com/downloads.htm>.

Other remarks: As shown in Table 1, all markers showed high PD values (>0.800) the highest being that observed for D2S1338 and lowest for TPOX. Marker D18S51 was the most powerful marker used for paternity testing with the highest PE and TPI. Deviation from Hardy–Weinberg equilibrium was observed for D21S11, however this deviation was not observed after correction using FSTAT ($p = 0.667$; 2000 randomisations).

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