



POP-Gene TIMOR: first forensic DNA marker study of East-Timor people

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Abstract. The first population-genetics study for the world newest independent country, East Timor, is presented. In this preliminary work, part of a major ongoing study on East-Timor genetic diversity, the allele frequencies and some statistical parameters of forensic interest were determined for the 15 loci included in AmpFLSTR Identifiler™ genotyping kit. A total of 107 samples, collected from East Timorese of several linguistic groups, was typed. All markers are in Hardy-Weinberg equilibrium (except for D2S1338 and D5S818, but the deviations do not reach significance after Bonferroni correction). Observed heterozygosities varied between 72% (D5S818) and 92% (D8S1179). © 2003 Elsevier B.V. All rights reserved.

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1. Introduction

East Timor, a Portuguese colony from the 16th century until Indonesian occupation in 1975, gained finally its independence, after a tragic and complex political process on May 20th, 2002. East Timor is indeed a complex multilingual mosaic and its ethnic and linguistic heterogeneity has long attracted scientific interest in areas such as anthropology, linguistics and archaeology and constitutes a challenge to human genetics.

The AmpFLSTR® Identifiler™ PCR Amplification Kit (*Applied Biosystems*) is being widely used in forensics (as well as in paternity studies), as it complies with international standards and recommendations such as those from the *Combined DNA Index System* (CODIS), the *European Network of Forensic Science Institutes* (ENFSI) and Interpol. We present preliminary data from the first (to our knowledge) DNA study concerning East-Timorese populations, as part of a broad ongoing study of East-Timor

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Table 1
Allele frequencies and statistical parameters of forensic relevance, for the 15 Identifier™ STRs in East-Timor population

	D8S1179	D21S11	D7S820	CSF1PO	D3S1358	HUMTH01	D13S317	D16S539	D2S1338	D19S433	HUMVWA	TPOX	D18S51	D5S818	FGA
6						0.098						0.005			
7			0.009			0.159									
8	0.005		0.257	0.005		0.350	0.299	0.028				0.332			
9			0.028	0.061		0.341	0.075	0.154				0.308		0.023	
9.3						0.009									
10	0.042		0.229	0.210		0.042	0.112	0.210				0.028		0.318	
11	0.126		0.285	0.332			0.248	0.318				0.299	0.005	0.229	
12	0.112		0.168	0.290	0.005		0.201	0.182		0.047		0.028	0.037	0.290	
13	0.201		0.019	0.093	0.005		0.051	0.093		0.308			0.079	0.136	
13.2										0.023					
14	0.234		0.005	0.009	0.009		0.014	0.009		0.243	0.117		0.196	0.005	
14.2										0.075			0.005		
15	0.112				0.336			0.005		0.056	0.084		0.145		
15.2										0.182					
16	0.098				0.322					0.033	0.173		0.150		
16.2										0.028					
17	0.065				0.266				0.089	0.005	0.243		0.178		
18	0.005				0.051				0.042		0.257		0.070		0.005
19					0.005				0.215		0.117		0.056		0.089
20									0.042		0.009		0.019		0.051
21									0.028				0.028		0.117
22									0.107				0.019		0.206
23									0.206				0.005		0.206
24									0.192						0.154
25									0.070				0.005		0.093
26									0.005				0.005		0.047
27		0.005							0.005						0.028
28		0.112													0.005
29		0.262													
30		0.196													
30.2		0.005													
31		0.103													
31.2		0.103													
32		0.019													
32.2		0.154													
33		0.005													
33.2		0.028													
35.2		0.005													
37.2		0.005													
H obs.	0.91589	0.84112	0.80374	0.76636	0.74766	0.74766	0.82243	0.79439	0.76636	0.85981	0.84112	0.77570	0.86916	0.71963	0.90654
H exp.	0.85222	0.83818	0.77434	0.75275	0.71541	0.72748	0.79154	0.79356	0.85007	0.80628	0.81436	0.70708	0.87333	0.74722	0.85977
P	0.25516	0.53635	0.20094	0.69158	0.36875	0.26378	0.79872	0.75580	0.02591	0.80129	0.14264	0.20487	0.46464	0.01221	0.36966

H obs.: Observed Heterozygosity; H exp.: Expected Heterozygosity.

P: Hardy-Weinberg equilibrium P value, exact test based on more than 2000 shuffling, for standard error <0.01.

Genetics (*POP-Gene TIMOR*), which involves the cooperative efforts of the Ministry of Health of the *República Democrática de Timor-Leste* (RDTL) and Portuguese institutions.

2. Materials and methods

Blood/saliva samples, in FTA™ cards, were collected from 107 unrelated donors (with informed consent), belonging to different geographic (and linguistic) origins in East Timor. The DNA was extracted using the Chelex procedure [1] over a 3-mm² cut of the FTA™ card.

Amplification of the 15 STR Identifiler™ loci (CSF1P0, D2S1338, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D19S433, D21S11, FGA, TH01, TPOX, VWA), plus the gender marker Amelogenin, was performed following the manufacturer's recommendations.

Amplified fragments were separated using an ABI 310 Automated Sequencer and analysed with GeneScan Software. All “off-ladder” fragments were typed at least twice. The population-genetics analyses were performed using the Arlequin software package [2].

3. Results and discussion

Allele frequencies (and relevant forensic statistical parameters) for the East-Timor subjects studied are shown in Table 1 for the 15 Identifiler™ STRs. No deviations from H–W equilibrium were observed with the exception of D2S1338 and D5S818 loci, but the deviations do not reach significance after Bonferroni correction. The observed heterozygosities varied between 72% (D5S818) and 92% (D8S1179) and the observed average heterozygosity (81%) is not significantly different from the expected value (79%).

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