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Article *in* Forensic Science International: Genetics · April 2009

DOI: 10.1016/j.fsigen.2008.05.007 · Source: PubMed

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Genetic variation of 15 autosomal STR loci in Upper (Southern) Egyptians

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Received 1 March 2008; accepted 12 May 2008

Abstract

A sample of 265 unrelated individuals inhabiting five governorates in Upper (south) Egypt was collected with informed consent. The samples were amplified using the AmpFℓSTR® Identifiler™ PCR Amplification Kit (containing 15 loci: D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, vWA, TPOX, D18S51, D5S818 and FGA), and genotyped subsequent to capillary electrophoresis. Statistical analysis of the generated data indicated neither departure from expectation of Hardy–Weinberg Equilibrium (HWE) in most of the tested loci nor dependence of alleles between loci. All tested loci were polymorphic; the most discriminating is D18S51 while the least is TPOX. The combined power of exclusion was 0.99999868 and the combined match probability was 1.93×10^{-18}. The genetic diversity of the Upper Egyptians was compared with those of other populations at the local, regional and global levels.

Keywords: Autosomal STRs; Population database; Upper Egyptians; South Egypt

Population: A sample of 265 unrelated volunteers from five governorates (El-Minia, Assiut, Sohag, Qena and Aswan) in Upper (south) Egypt (Fig. 1), was collected. After obtaining appropriate ethical approval, the participants were questioned for their ethnic origin as inhabitants of Upper Egypt for at least the third generation and they signed informed consents. The residents are culturally and linguistically distinguishable from those of the Northern population, where people speak an Arabic language with a distinctive dialect.

Extraction: Genomic DNA was extracted from blood stains and buccal swabs by a modified silica-based method [1]. DNA quantification was undertaken using the Quant-iT™ High Sensitivity DNA Assay Kit (Molecular Probes) according to the manufacturer's instructions.

PCR: 0.5–1 ng DNA templates were amplified in half quantities according to the manufacturer's instructions (AmpFℓSTR® Identifiler™ PCR Amplification Kit: User's Manual, Applied Biosystems).

Typing: The PCR products were detected with the ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems) and sized with GeneScan-500 LIZ internal lane size standard. Allelic calls and genotyping were carried out by comparison to the reference allelic ladder included in the kit, using GeneMapper ID® v3.2 (Applied Biosystems).

Quality control: Identifiler™ kit included allelic ladder and positive control.

Analysis of data: Microsoft Excel-PowerStats v1.2 [2] was used for allele frequency and forensic parameters calculations including heterozygosity (H), polymorphic information content (PIC), power of discrimination (PD), power of exclusion (PE) and matching probability (MP) for each locus of the studied population. Hardy–Weinberg equilibrium (HWE), Linkage disequilibrium (LD), population differentiation tests, *FST* genetic distances and pair-wise analysis of molecular variance (AMOVA) for comparison with other populations, were carried out using Arlequin v3.01 [3] and GenAlEx 6 software [4]. In addition, graphical representation of genetic distances (*FST* distances) using a two-dimensional genetic map based on multi-dimensional scaling (MDS) analysis was performed by means of SPSS v14.0 software. Both frequency data gathered from literature

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doi:10.1016/j.fsigen.2008.05.007

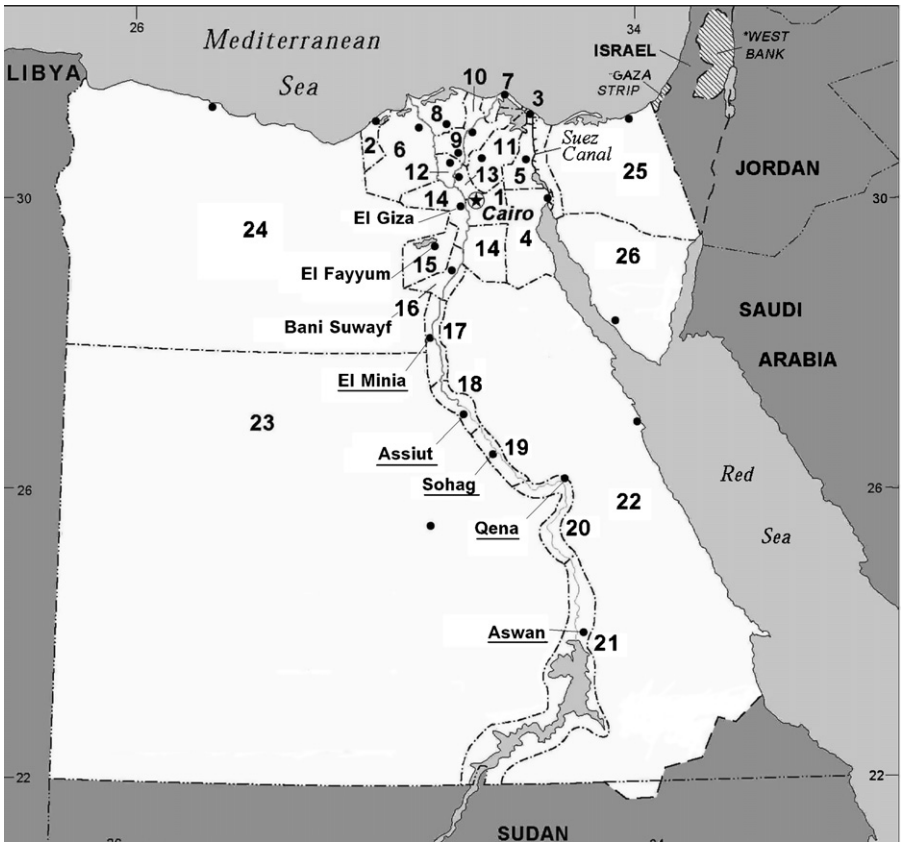

Fig. 1. Map of Egypt (26 governorates), showing the geographical location of Upper Egypt's governorates (14–21) with those included in the present study underlined; adapted from: http://lcweb2.loc.gov/frd/cs/egypt/eg00_06a.pdf.

and actual genotypic data obtained through personal communications, were considered for comparative analyses with other populations at the local, regional and global levels (Table 1) [5,6].

Access to the data: Supplementary data available online as an Excel file.

Results: Allele frequency data of the 15 STR loci are presented in Table 2. HWE evaluation and forensic parameters are included in Table 3. Assessment of genetic distances (*FST*) based on frequency data comparison with other local and regional population versus Upper Egyptians is collected in Table 4. Multi-dimensional scaling plot derived from the *FST* genetic distances among Upper Egyptians and other populations compared globally is shown in Fig. 2.

Other remarks: Pair-wise *FST* genetic distances between sampled subpopulations from Upper Egypt revealed no significant differences (data not shown), so they have been treated as a single population in subsequent analyses. The frequencies of alleles at all loci along with forensic parameters were addressed (Tables 2 and 3). All loci are highly polymorphic with the highest being D18S51 (0.88), and the lowest is TPOX (0.62). The most polymorphic loci were found to be the most discriminating as well. The combined power of exclusion was calculated to be 99.999868% and the combined

Table 1
Diverse (global) populations and constituent subpopulations included in the genetic distance analyses with Upper Egyptians

Population	Number	Code	Contained subpopulations
South Asian	198	SAS	Pakistani (Brahui, Balochi, Hazara, Makrani, Sindhi, Pathan, Kalash, Burusho)
East Asian	250	EAS	Japan, China (Han, Tuji, Yizu, Miaozu, Orogen, Daur, Mongolia, Hezhen, Xibo, Uygur, Dai, Lahu, She, Naxitu), Siberia (Yakut)
Sub-Saharan African	125	SSA	Central African Republic (Biaka), Congo (Mbuti), Senegal (Mandenka), Nigeria (Yoruba), Namibia (San), South Africa (Bantu), Kenya (Bantu)
European	158	EUR	France (Basque), Italy (Sardinian, Bergamo, Tuscan), Russia (Caucasus, Adygei), Orkney Islands
Oceanian	39	OCE	New Guinea (Papuan), Bougainville (Melanesian)
American	108	AM	Colombia, Brazil (Surui, Karitiana), Mexico (Maya, Pima)
North African	30	NAF	Algeria (Mozabites)
Middle Eastern	148	ME	Israel (Druze, Bedouin, Palestinian)

Table 2
Allele frequencies of 15 STR loci included in the Identifiler kit in Upper Egyptians

Allele	D8	D21	D7	CSFIPO	D3	TH01	D13	D16	D2	D19	vWA	TPOX	D18	D5	FGA
5			0.002												
6						0.206						0.009			
7			0.011	0.008		0.204						0.006			
8	0.015		0.16	0.021		0.115	0.126	0.036				**0.483**	0.002	0.051	
9	0.006		0.094	0.015		**0.374**	0.094	0.151				0.211		0.081	
9.3						0.081									
10	0.057		**0.342**	0.264		0.019	0.047	0.104		0.008		0.062	0.002	0.125	
10.2													0.002		
11	0.108		0.245	0.311		0.002	0.226	**0.268**		0.011		0.217	0.008	0.225	
11.2										0.002			0.009		
12	0.111		0.123	**0.338**			**0.338**	0.255		0.113		0.011	**0.166**	**0.364**	
12.2										0.008			0.004		
13	0.208		0.021	0.036	0.006		0.119	0.162		0.228	0.002		0.104	0.145	
13.2										0.034			0.002		
14	**0.242**		0.002	0.008	0.072		0.047	0.025		**0.247**	0.087		0.128	0.009	
14.2										0.053			0.002		
15	0.191				**0.306**		0.002			0.143	0.104		0.128		
15.2										0.057			0.002		
16	0.046				0.262				0.04	0.053	**0.292**		0.134		
16.1															0.002
16.2										0.04			0.002		
17	0.011				0.228				**0.232**	0.004	0.272		0.102		0.002
17.2													0.002		
18	0.004				0.117				0.136		0.164		0.074		0.011
18.2													0.002		
19					0.009				0.113		0.066		0.062		0.049
20									0.151		0.013		0.03		0.077
20.2															0.004
21									0.062				0.026		0.134
21.2															0.004
22									0.062				0.008		**0.192**
22.2															0.006
22.3															0.002
23									0.092						0.158
23.2															0.002
24									0.058						0.157
25									0.043						0.098
26		0.004							0.008						0.057
27		0.036							0.002						0.023
28		0.123													0.015
28.2		0.004													
29		**0.268**													0.006
29.2		0.002													
30		0.243													0.002
30.2		0.013													
31		0.047													
31.2		0.089													
32		0.004													
32.2		0.106													
33		0.002													
33.2		0.04													
34		0.004													
34.2		0.006													
35		0.006													
36		0.002													
37		0.002													
38		0.002													

The highest allele frequency for each locus is in bold.

match probability was 1.93×10^{-18}. Hardy–Weinberg Equilibrium expectations were met for all loci except for D18S51, D5S818 and D19S51 (p-values: 0.000, 0.000 and 0.013, respectively) (Table 3). However, the exact test p-values for the first two loci remained significant even after Bonferroni's correction. Possible explanations for HWE departure are population substructure and admixture, since there is persistent consanguinity (27.7% in WHO report, 1994) [7] in addition to

Table 3
Forensic parameters and the exact test probability values for Hardy–Weinberg equilibrium evaluation

Locus	D8	D21	D7	CSFIPO	D3	TH01	D13	D16	D2	D19	vWA	TPOX	D18	D5	FGA
MP	0.05	0.051	0.087	0.133	0.1	0.096	0.075	0.069	0.033	0.045	0.076	0.16	0.026	0.09	0.033
PEX	0.627	0.693	0.558	0.473	0.565	0.431	0.524	0.592	0.753	0.565	0.48	0.37	0.664	0.551	0.807
PD	0.95	0.949	0.913	0.867	0.9	0.904	0.925	0.931	0.967	0.955	0.924	0.84	0.974	0.91	0.967
PIC	0.81	0.81	0.74	0.66	0.73	0.73	0.76	0.77	0.86	0.82	0.76	0.62	0.88	0.74	0.86
H (%)	81.5	84.9	77.7	72.5	78.1	70.2	75.8	79.6	87.9	78.1	73.2	66	83.4	77.4	90.6
Ho	0.815	0.849	0.777	0.728	0.781	0.706	0.758	0.796	0.879	0.781	0.732	0.660	0.834	0.774	0.906
He	0.834	0.831	0.777	0.720	0.768	0.757	0.794	0.805	0.871	0.844	0.793	0.673	0.891	0.795	0.874
p	0.165	0.947	0.489	0.219	0.105	0.182	0.337	0.459	0.861	**0.013**	0.119	0.414	**0.000**	**0.000**	0.341

MP, match probability; PEX, power of exclusion; PD, power of discrimination; PIC, polymorphism information content; H, heterozygosity; Ho, heterozygosity observed; He, heterozygosity expected. Significant *p* values are in bold.

historical admixture with other ethnic groups who ruled Egypt in its past, e.g. Ottomans, Arabs (who constituted major migration waves) and Greeks. There was no evidence of departure from independence of alleles between all tested loci (linkage equilibrium).

Local comparisons between Upper Egyptians were carried out with other ethnic groups in Egypt, based on frequency and molecular data. No differences were observed in comparison with a general Caucasian population from Cairo in any of the nine loci compared [8] or with Egyptian Christians from Cairo [9], but one out of eight loci showed a difference in comparison with a population from El-Minia city [10]. At the molecular data level, there was a weak significant difference when Upper Egyptians were compared with Egyptian Muslims from Tanta (*FST* *p*-value = 0.0455), albeit with a non-significant *p*-value (0.143) in an exact test of population differentiation. Highly significant differences ($p < 0.00001$) were observed in comparisons with Berbers from Siwa (an oasis in the western desert), and with a population sample from Adaima (a village in Upper Egypt) [11].

Global and regional comparisons, based on genotypes of all loci except D2S1338 and D19S433, with diverse populations from South Asia, sub-Saharan Africa, Europe, Oceania, Middle East, the Americas and North Africa were performed to address relationships with neighbouring and distant populations. A highly significant *FST* genetic difference was observed with all groups (*p*-values <0.00001). The genetic relationships with these populations were demonstrated in a multi-dimensional scaling plot which showed that Upper Egyptians are located centrally in relation to other continental groups. They were very distant from Oceanian and American populations and clustered with Middle Eastern populations (including Israeli Druze, Bedouins and Palestinians) rather than with Europeans, South Asians and North Africans (Fig. 2).

Further comparisons of genetic distances were undertaken with other related Arab and European populations by means of published allele frequency data [12–15] (Table 4). These showed the following significant differences: 5 out of 15 loci in Qatari; three and one out of 13 loci in Syrians and Moroccans, respectively; 1 and 4 out of 12 loci in Algerian Mozabites and Tunisians, respectively; 1 out of 9 loci in Omani; 2 out of 7 in an Israeli Jewish sample. No difference was observed with Yemenites in the seven loci examined. Regarding the European populations, Turkish, Greeks and Italians, significant dissimilarity was found in 7 out of 15 loci, 1 out of 9 and 4 out of 13, respectively [16–18].

Clustering of Upper Egyptians with Middle Eastern populations, and weak differentiation from most other Arab populations, is expected because of geographical proximity and gene flow to Egypt, which has acted as a crossroad to the three continents throughout its history. This observation is supported by mitochondrial DNA and Y chromosome studies which have suggested a regional genetic continuity among populations of the Nile valley, Middle East and Arabian Peninsula [19,20]. This paper follows the guidelines suggested for publication of population data [21].

Acknowledgments

We thank all DNA donors, Emma Parkin for technical support, and Patricia Balaresque for advice on statistical analysis. We also thank Peter de Knijff for supplying

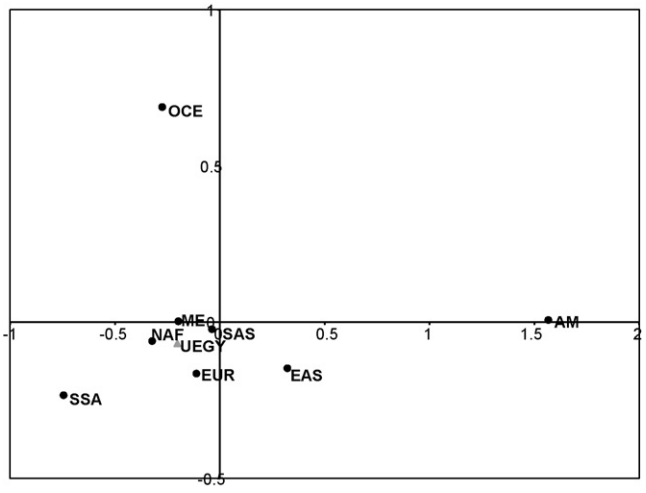

Fig. 2. Multi-dimensional scaling (MDS) based on pair-wise *FST* genetic distances of Upper Egyptian and other diverse global populations. OCE, Oceanian; ME, Middle Eastern; NAF, North African; EAS, East Asian; SSA, sub-Saharan African; UEGY, Upper Egyptian; SAS, South Asian; EUR, European. The figure shows that Oceania and American populations are very distant from Upper Egyptians (marked by a grey triangle) and other populations. The Upper Egyptian population is closer to the Middle Eastern, North African, South Asian and European populations than others.

Table 4
Genetic distances (*FST*) *p*-values between Upper Egyptians and other related populations.

Loci	U.Eg vs. Eg.El-Minia	U.Eg vs. Eg.Christians	U.Eg vs. Eg. Cairo	U.Eg vs. Dubai	U.Eg vs. Is. Jewish	U.Eg vs. Is. Arabs	U.Eg vs. Syrian	U.Eg vs. Moroccan	U.Eg vs. Qatari	U.Eg vs. Omani	U.Eg vs. Tunisean	U.Eg vs. Algerian	U.Eg vs. Yemenite	U.Eg vs. Turkish	U.Eg vs. Greek	U.Eg vs. Italian (NW)
D8S117	–	0.578	0.991	0.431	–	–	0.207	0.668	0.334	0.476	0.199	0.142	0.145	**0.019**	0.847	**0.000**
D21S11	–	0.849	0.358	0.074	–	–	0.570	0.811	0.636	0.209	0.991	0.617	–	0.537	0.674	0.701
D7S820	0.907	–	0.180	0.950	0.189	0.153	0.936	0.199	0.579	0.999	0.828	0.567	0.939	**0.038**	0.513	0.567
CSFIPO	0.456	–	–	0.263	0.377	0.352	0.622	0.994	0.988	–	**0.000**	0.605	–	0.853	–	0.721
D3S1358	0.672	0.729	0.769	0.516	–	–	0.651	0.826	0.147	**0.000**	0.893	0.808	0.179	0.475	0.080	0.422
TH01	**0.001**	–	–	**0.000**	**0.000**	**0.000**	**0.028**	0.336	**0.045**	–	**0.001**	0.439	–	**0.000**	–	**0.000**
D13S317	O.2C0	0.991	0.447	0.255	**0.011**	0.736	0.056	0.217	0.102	0.440	**0.008**	0.499	0.283	**0.007**	0.145	0.493
D16S539	0.633	–	–	0.571	0.219	0.183	0.914	0.433	**0.004**	–	0.298	–	–	0.584	–	0.C97
D2S1338	–	–	–	–	–	–	–	–	0.857	–	–	–	–	0.719	–	–
D19S433	–	–	–	–	–	–	–	–	**0.023**	–	–	–	–	0.216	–	–
vWA	0.368	0.685	0.968	0.434	0.121	0.895	0.726	0.508	0.567	0.668	0.419	0.142	0.647	0.056	**0.045**	**0.025**
TPOX	0.960	–	–	0.086	0.061	0.155	**0.000**	**0.000**	**0.000**	–	**0.000**	**0.000**	–	**0.000**	–	**0.000**
D18S51	–	0.977	0.192	0.336	–	–	0.441	0.527	0.071	0.341	0.267	0.638	–	**0.022**	0.373	0.871
D5S818	0.535	0.592	0.288	0.153	–	–	**0.029**	0.164	0.134	0.372	0.128	0.169	0.793	**0.032**	0.307	0.129
FGA	0.934	0.449	0.223	0.808	–	–	0.249	0.832	**0.000**	0.823	0.763	0.052	0.643	0.252	0.984	0.091

Numbers in bold indicate significant (*FST*) *p*-values (significance level is 0.05). U.Eg, Upper Egyptians; IS, Israeli; NW, North West.

unpublished comparative data on the CEPH-HGDP diversity panel. We are grateful to the Egyptian government for funding this research. Mark A. Jobling is a Wellcome Trust Senior Fellow in Basic Biomedical Science (grant number 057559).

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2008.05.007.

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