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SHORT COMMUNICATION

Meiosis study in a population sample from Nigeria: allele frequencies and mutation rates of 16 STR loci

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Abstract Allele frequencies for the 16 short tandem repeat (STR) loci D2S1338, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D19S433, D21S11, ACTBP2, CSF1PO, FGA, TH01, TPOX and VWA were determined for 337 immigrants from Nigeria. All loci were in Hardy–Weinberg equilibrium. More than 6,000 meiotic transfers were investigated and ten mutations were observed. Single mutations were observed in the STR systems D2S1338, D3S1358, D7S820, D8S1179, D16S539 and FGA, whereas two mutations were observed in the systems D21S11 and VWA.

Keywords Short tandem repeat (STR) · Nigeria · Population study · Mutation

Introduction

To increase our knowledge of Sub-Saharan African populations, we have investigated 16 short tandem repeat (STR) systems (i.e. D2S1338, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D19S433, D21S11, ACTBP2, CSF1PO, FGA, TH01, TPOX and VWA) in 337 unrelated Nigerian immigrants, 150 women and 187 men, seeking asylum in Germany. In this paper, we present the allele frequencies, mutation rates and forensic efficiency values for the analysed STR loci.

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Materials and methods

Genomic DNA was extracted from buccal swabs using a modified Chelex method [1]. Amplification was carried out using various kits (e.g. AmpF*l*STR Profiler, SEFiler, Identifiler PCR amplification kits from Applied Biosystems, Darmstadt, Germany and Power ES Kit from Promega, Mannheim, Germany). Polymerase chain reaction products were analysed on ABI PRISM 310 or 3100-Avant Genetic Analyzers according to the manufacturer's instructions.

The forensic efficiency data were calculated with the software HWE Analysis 3.2 (Chr. Puers, Münster). De novo mutations were characterised as described earlier [2].

Variant alleles and alleles from the mutation cases were isolated as described elsewhere [3] and directly sequenced using BigDye Terminator Cycle Sequencing Kit (ABI) with primers for both strands to check if the mutation had occurred in the repeat region.

Results and discussion

No deviation from Hardy–Weinberg equilibrium was observed for the 16 STR loci (Table S1 of the Electronic supplementary material). The most informative STR systems in the Nigerian population sample were ACTBP2, D18S51 and FGA with their power of discrimination ranging from 0.980 to 0.968 and their mean exclusion chance (MEC) ranging from 0.827 to 0.748. The combined matching probability for the 16 STR loci in the Nigerian population is 1 in $1{,}4\times10^{19}$ and the combined MEC is 0.99999992. According to these statistical parameters, the 16 STR loci are well-suited for parentage testing and forensic casework purposes in the Nigerian population.

Table 1 Characteristics of the ten mutation cases from Nigeria

Case no.	System	Child	Mother	Alleged father	Origin	Gain/loss	Sequence
E205/06	D2S1338	*19*/25	24/25	*20*/23	Paternal	−1	$(TGCC)_7$ $(TTCC)_{13\rightarrow12}$
E275/06	D3S1358	*16*/17	14/17	*17*/18	Paternal	−1	$(AGAT)_{14\rightarrow13}$ (AGAC) $(AGAT)_2$
E191/05	D7S820	10/*11*	10	8/*12*	Paternal	−1	$(GATA)_{12\rightarrow11}$
E360/02	D8S1179	15/*17*	10/15	15/*16*	Paternal	+1	$(TATC)_3$ TGTC $(TATC)_{12\rightarrow13}$
E118/04	D16S539	10/*12*	n.t.	*11*	Paternal	+1	$(GATA)_{11\rightarrow12}$
E003/04	D21S11	29/*33.2*	31.2/*34.2*	n.t.	Maternal	−1	$(TCTA)_5$ $(TCTG)_6$–$(TCTA)_{14\rightarrow13}$ TA TCTA
E290/01a	D21S11	30/*36*	29/30	28/*35*	Paternal	+1	$(TCTA)_9$ $(TCTG)_5$–$(TCTA)_{13\rightarrow14}$
E182/04	FGA	*21*/22	*22*	22/23	Unassigned	−1	$(TTTC)_3$ TTTT TTCT $(CTTT)_{14\rightarrow13}$ CTCC $(TTCC)_2$
E252/03a	VWA	16/*19*	16	15/*20*	Paternal	−1	TCTA $(TCTG)_4$ $(TCTA)_{15\rightarrow14}$
E081/05	VWA	*14*/18	n.t.	*13*/17	Paternal	+1	TCTA $(TCTG)_4$ $(TCTA)_{10\rightarrow11}$ $(TCCA)_2$ (TCCT)

Alleles affected by the mutational event are shown in italics
n.t. not available for typing
Constant region (43 bp): $(TCTA)_3$ TA $(TCTA)_3$ TCA $(TCTA)_2$ TCC ATA

In the immigration cases with highly validated parentage ($W \geq 99.99\%$), we have observed a total of ten one-step mutations under 3,023 maternal and 3,273 paternal meiotic transfers, eight in the male and one in the female germ line, while one mutation could not be assigned. Repeat contractions were observed in six cases, repeat expansions in four cases (Table 1). The observed mutation rates ranged from 0 to 0.99×10^{-2} per locus per gamete per generation (Table 2) and, thus, are in the range reported by Brinkmann et al. [4]. A correlation between the mutation rate and age of parents could not be observed (Table S2 of the Electronic supplementary material), but the number of Nigerian STR mutations is yet too small to draw statistically significant conclusions.

Sequencing assisted to properly characterise the mutation in three cases (Table S3 of the Electronic supplementary material), either by differences in the flanking regions or by the presence of iso-alleles, e.g. in case E275/06, the filial allele 16 might have originated from the paternal or maternal allele 17. The sequence structure of the filial allele 16 showed the repeat motif $(AGAT)_{13}$ (AGAC) $(AGAT)_2$ which is compatible with the paternal allele 17: $(AGAT)_{14}$ (AGAC) $(AGAT)_2$ but incompatible with the maternal allele 17: $(AGAT)_{12}$ $(AGAC)_3$ $(AGAT)_2$. Thus, this mutation could be classified as a paternal one-step loss.

In the deficiency case E118/04, the filial allele 10 or 12 might have originated from the paternal allele 11. Sequencing of the filial alleles revealed the motif $(GATA)_{10}$ and

Table 2 Mutation rates of the 16 STR systems in Nigeria

System	Paternal mutations	Maternal mutations	Unassigned	Paternal transmissions	Paternal mutation rate (%)	95% confidence interval (%)	Maternal transmissions	Maternal mutation rate (%)	95% confidence interval (%)
D2S1338	1			101	0.99	0.17–5.40	132	0	0.00–2.83
D3S1358	1			237	0.42	0.07–2.35	247	0	0.00–1.53
D5S818				237	0.00	0.00–1.60	245	0	0.00–1.54
D7S820	1			234	0.43	0.08–2.38	244	0	0.00–1.55
D8S1179	1			160	0.63	0.11–3.46	180	0	0.00–2.09
D13S317				237	0.00	0.00–1.60	247	0	0.00–1.53
D16S539	1			101	0.99	0.17–5.40	132	0	0.00–2.83
D18S51				158	0.00	0.00–2.37	177	0	0.00–2.12
D19S433				101	0.00	0.00–3.66	132	0	0.00–2.83
D21S11	1	1		160	0.63	0.11–3.46	180	0.56	0.10–3.09
ACTBP2				113	0.00	0.00–3.29	129	0	0.00–2.89
CSF1PO				236	0.00	0.00–1.60	246	0	0.00–1.54
FGA			1	237	0.00	0.00–1.60	245	0	0.00–1.54
TH01				237	0.00	0.00–1.60	247	0	0.00–1.54
TPOX				237	0.00	0.00–1.60	245	0	0.00–1.54
VWA	2			237	0.84	0.23–3.02	245	0	0.00–1.54
Total	8	1	1	3023			3273		

The 95% confidence interval was online-calculated (http://faculty.vassar.edu/lowry/prop1.html)

$(GATA)_{12}$, whereas the paternal allele 11 showed the motif $(GATA)_{11}$. Additional information was obtained from the flanking region: the filial allele 10 revealed a base substitution from A to C 16 bp upstream of the repeat region, whereas the filial allele 10 and the paternal allele 11 showed the regular sequence structure. Thus, this mutation could be classified as a paternal one-step gain.

Some rare alleles observed in the Nigerian population were sequenced (Table S4 of the Electronic supplementary material) and the following novel sequences were found: alleles 23.3 and 45.2 at FGA, alleles 7.3 and 10.3 at D7S820 and allele 33.3 at D21S11.

To conclude, we have established a forensic database for allele frequencies and mutation rates in the Nigerian population which is useful for identity and parentage testing.

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