

## SHORT COMMUNICATION

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## The Bubi population of Equatorial Guinea characterised by HUMTH01, HUMVWA31A, HUMCSF1PO, HUMTPOX, D3S1358, D8S1179, D18S51 and D19S253 STR polymorphisms

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**Abstract** Allele frequencies for eight STR loci (HUMTH01, HUMVWA31A, HUMCSF1PO, HUMTPOX, D3S1358, D8S1179, D18S51, D19S253) have been analysed in the Bubi population of Bioko Island, Equatorial Guinea. For all loci, no deviation from Hardy-Weinberg equilibrium was found. Data obtained were compared with that of Caucasian and African populations. Significant differences were found for all systems between all the black populations compared and the Caucasoid population. Similarities were observed between the Bubi and Zimbabweans, and also with African American populations. Also, more affinities were observed between Zimbabweans and Ugandans and Ovambos than between these groups and the Bubi population. From these comparisons it is suggested that in Africa, as in other continents, there is a certain genetic heterogeneity.

**Keywords** HUMTH01 · HUMVWA31A · HUMCSF1PO · HUMTPOX · D3S1358 · D8S1179 · D18S51 · D19S253 · Africa · Bubi population

### Introduction

Short tandem repeat (STR) polymorphisms are not only useful for medico-legal studies but also for genetic analyses of populations. In this sense, the available data for a large number of polymorphisms, for example of black

populations, are extremely generalised and as a consequence, it is difficult to know their variability as with other human populations. The purpose of this work was to study eight STR polymorphisms in an African population such as the Bubis from Bioko Island (Equatorial Guinea).

The Bubi are an old Bantu people and form the major indigenous group on Bioko Island, previously called Fernando Poo, a volcanic island in the Bight of Biafra about 30 km from continental Equatorial Guinea, West Central Africa.

### Material and methods

DNA was extracted from blood stain samples from healthy unrelated Bubi individuals using Chelex 100 and the method described by Walsh et al. [1]. PCR triplex (D3S1358, D18S51, D19S253 and HUMTH01, HUMVWA31A, HUMCSF1PO, HUMTPOX) and singleplex (D8S1179, HUMVWA31A) amplifications were accomplished with fluorescein-labelled primers by the methods described by Gené et al. [2, 3, 4]. Genotypes were analysed in denaturing 6% polyacrylamide gel electrophoresis, using a monochrome automated laser fluorescence sequencer (ALF Pharmacia, Uppsala). For all markers, possible divergences from Hardy-Weinberg equilibrium (HWE) were determined by calculating the exact test proposed by Guo and Thompson [5]. The Bubi data were compared with African populations using a  $R \times C$  contingency table  $\chi^2$  test for homogeneity [6]. From a forensic point of view, the heterozygosity value ( $h$ ) [7], the power of discrimination (PD) [8], and the a priori chance exclusion value (CE) [9] were calculated.

### Results and discussion

The number of genotypes and alleles observed in each system and the allele frequencies obtained for the eight STR loci in the Bubi population are shown in Table 1. No significant deviations from Hardy-Weinberg expectations were found for all STRs. The data obtained for the eight loci were compared with that of African populations such as Zimbabwe [10], Ovambos from Namibia and Ugandans [11], and African Americans from USA [12]. Except for Zimbabwe, the rest of the groups can only be compared using two or three polymorphisms. Significant dif-

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**Table 1** Allele frequency distributions for HUMTH01, HUMVWA31A, HUMCSF1PO, HUMTPOX, D3S1358, D8S1179, D18S51 and D19S253 in the Bubi population

Allele	HUMTH01 (n = 151)	HUMVWA31A (n = 130)	HUMCSF1PO (n = 149)	HUMTPOX (n = 155)	D3S1358 (n = 148)	D8S1179 (n = 123)	D18S51 (n = 125)	D19S253 (n = 142)
3	—	—	—	—	—	—	—	—
5	—	—	—	—	—	—	—	—
6	0.113	—	—	0.116	—	—	—	0.028
7	0.437	—	0.074	0.029	—	—	—	0.194
8	0.261	—	0.047	0.258	—	—	—	0.046
9	0.156	—	0.037	0.232	—	—	0.020	0.099
9.3	0.003	—	—	—	—	—	—	—
10	0.030	—	0.275	0.103	—	0.004	—	0.060
11	—	0.015	0.221	0.255	—	0.033	—	0.180
12	—	—	0.305	0.006	0.020	0.183	0.028	0.225
13	—	0.011	0.037	—	0.003	0.187	0.024	0.130
14	—	0.042	0.003	—	0.088	0.382	0.116	0.035
15	—	0.273	—	—	0.250	0.150	0.148	0.003
16	—	0.296	—	—	0.375	0.057	0.160	—
17	—	0.188	—	—	0.236	0.004	0.204	—
18	—	0.108	—	—	0.027	—	0.112	—
19	—	0.050	—	—	—	—	0.124	—
20	—	0.015	—	—	—	—	0.016	—
21	—	—	—	—	—	—	0.028	—
22	—	—	—	—	—	—	0.012	—
42	—	—	—	—	—	—	0.008	—
HWE exact test	$P = 0.6437$	$P = 0.2687$	$P = 0.6521$	$P = 0.5484$	$P = 0.1827$	$P = 0.1074$	$P = 0.1717$	$P = 0.4323$

**Table 2** African populations compared for HUMTH01, HUMVWA, HUMCSF1PO and HUMTPOX systems

System	Populations compared	Probability
HUMTH01	Bubi vs Zimbabweans	$P = 0.5462$
	Bubi vs Ugandans	$P = 1.304\text{E-}03$
	Bubi vs Ovambos	$P = 7.450\text{E-}03$
	Bubi vs African Americans	$P = 4.874\text{E-}07$
	Zimbabweans vs Ugandans	$P = 3.823\text{E-}03$
	Zimbabweans vs Ovambos	$P = 0.1567$
	Zimbabweans vs African Americans	$P = 2.648\text{E-}04$
	Ugandans vs Ovambos	$P = 7.780\text{E-}08$
	Ugandans vs African Americans	$P = 9.027\text{E-}03$
	Ovambos vs African Americans	$P = 2.540\text{E-}07$
HUMVWA	Bubi vs Zimbabweans	$P = 3.114\text{E-}03$
	Bubi vs Ugandans	$P = 2.101\text{E-}03$
	Bubi vs Ovambos	$P = 0.0321$
	Zimbabweans vs Ugandans	$P = 0.4455$
	Zimbabweans vs Ovambos	$P = 1.212\text{E-}03$
	Ugandans vs Ovambos	$P = 6.846\text{E-}03$
HUMCSF1PO	Bubi vs Zimbabweans	$P = 0.2613$
	Bubi vs African Americans	$P = 0.3145$
	Zimbabweans vs African Americans	$P = 0.5855$
HUMTPOX	Bubi vs Zimbabweans	$P = 0.035$
	Bubi vs African Americans	$P = 0.0235$
	Zimbabweans vs African Americans	$P = 0.1985$
D3SS1358	Bubi vs Zimbabweans	$P = 0.7021$

**Table 3** Statistical parameters of forensic interest ( $h$  heterozygosity value, PD power of discrimination, CE chance of exclusion)

System	h	PD	CE
HUMTH01	0.705	0.865	0.462
HUMVWA31A	0.789	0.923	0.586
HUMCSF1PO	0.774	0.912	0.561
HUMTPOX	0.792	0.923	0.585
D3S1358	0.740	0.886	0.501
D8S1179	0.762	0.908	0.550
D18S51	0.870	0.968	0.731
D19S253	0.848	0.957	0.691

ferences were observed for all systems in the comparisons between all the black populations and the Caucasoid population studied by us [2, 3, 4]. To simplify, in Table 2 we show the comparisons between black populations and the probability of significance and it can be deduced that the Bubi population has a great affinity with the Zimbabwe sample. Also, the Bubi population has some similarities with the African Americans from USA. This result is in agreement with that observed by Budowle et al. in Zimbabweans [10]. In contrast, significant differences were observed between Ovambos and Ugandans, as well as between these populations and African Americans. No differences existed for HUMTH01 between Zimbabweans and Ovambos, and for HUMVWA between Zimbabweans and Ugandans. In general there seem to be more affinities between Zimbabweans and Ugandans and Ovambos than between these groups and the Bubi population. These re-

sults suggest that in Africa, as in other continents, there is a certain genetic heterogeneity. However, in order to draw more conclusions it would be necessary to analyse more populations and more polymorphisms.

The expected heterozygosity and the power of discrimination calculated from the gene frequencies obtained in our population (Table 3) reveal that in combination the eight systems have a high forensic efficiency.

## References

- Walsh PS, Metzger DA, Higuchi R (1991) Chelex 100 as a medium for simple extraction of DNA for PCR-based typing from forensic material. *Biotechniques* 10: 506–513
- Gené M, Huguet E, Moreno P, Sánchez C, Carracedo A, Corbella J (1996) Population study of the STRs HUMTH01 (including a new variant) and HUMVWA31A in Catalonia (northeast Spain). *Int J Legal Med* 108: 318–320
- Gené M, Carracedo A, Huguet E, Pérez-Pérez A, Moreno P (1998) Population genetics of the D12S391, CSF1PO and TPOX loci in Catalonia (northeast Spain). *Int J Legal Med* 111: 52–54
- Gené M, Piqué E, Borrego N, Carracedo A, Huguet E, Moreno P (1998) Catalonian population study of the tetranucleotide repeat loci D3S1358, D8S1179, D18S51 and D19S253. *Int J Legal Med* 112: 75–77
- Guo SW, Thompson EA (1992) Performing the exact test of Hardy-Weinberg proportion for multiple alleles. *Biometrics* 48: 361–372
- Fisher RA (1954) Statistical methods for research workers. Oliver and Boyd, Edinburgh
- Nei M, Roychoudhury AK (1974) Sampling variances of heterozygosity and genetic distance. *Genetics* 76: 379–390
- Fisher RA (1951) Standard calculations for evaluating a blood group system. *Heredity* 5: 95–102
- Ohno Y, Sebetan MI, Akaishi S (1982) A simple method for calculating the probability of excluding paternity with any number of codominant alleles. *Forensic Sci Int* 19: 93–98
- Budowle B, Nhari LT, Moretti TR, Kanoyangwa SB, Masuka E, Defenbaugh DA, Smerick JB (1997) Zimbabwe black population data on the six short tandem repeat loci – CSF1PO, TPOX, TH01, D3S1358, VWA and FGA. *Forensic Sci Int* 90: 215–221
- Brinkmann B, Junge A, Meyer E, Wiegand P (1998) Population genetic diversity in relation to microsatellite heterogeneity. *Hum Mutat* 11: 135–144
- Destro-Bisol G, Maviglia R, Caglià A, Boschi I, Spedini G, Pascali V, Clark A, Tishkoff S (1999) Estimating European admixture in African Americans by using microsatellites and a microsatellite haplotype (CD4/Alu). *Hum Genet* 104: 149–157