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Forensic parameters and genetic structure based on Y-chromosome short tandem repeats in Lesotho populations

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ABSTRACT

Lesotho is a landlocked country with approximately 2.2 million inhabitants. Over 97% of the population is represented by the Southern Sotho people (Sotho-Tswana group), followed by a number of minorities mostly from the Nguni language group. In this study we investigated the patterns of genetic variation and report genetic diversity, forensic parameters and novel allele variations in 938 unrelated Bantu males. Population pairwise comparisons identified high affinities between the Xhosa and the Vundle, while the largest genetic distance was observed between the Vundle and the Baphuthi ethnic groups (R_{st} = 0.14878). A high level of genetic differentiation between populations was observed considering culture and language affiliations as opposed to geographic distance.

1. Introduction

An average of 1556 rape cases per year was reported to the Lesotho Police from 2009 to 2016 (Unpublished data). Adoption of forensic kits targeting male-specific Y-chromosome short tandem repeats (Y-STR) proved useful in forensic investigations of sexual assault cases [1]. The application of forensic kits requires population-specific reference data to evaluate the significance of a DNA match. In this work, we generated and evaluated Lesotho population data with the UniQ Typer™ Y-10 panel, designed in South Africa [2], which was highly informative in South African Bantu populations [3,4].

2. Material and methods

Ethical clearance was granted by the Lesotho Ministry of Health and Social Welfare (ID128-2016) and the University of the Western Cape Senate Research Committee (15/4/97). DNA was extracted from 938 saliva samples collected [5] from unrelated males across the five Bantu ethnic groups residing the 10 Lesotho districts constituting three regions; North, Central and South and amplified with the UniQ Typer™ Y-10 genotyping kit [4,6]. Haplotype frequencies and Discrimination Capacity (DC) were computed using the counting method in ARLEQUIN v 3.5.2.2 [7]; Random Match Probability (RMP) was estimated as in [8], and Haplotype diversity (HD) as in [9]. Population genetic structure and the pairwise genetic distances (R_{st}) were analysed for 874 individuals (excluding duplications and nulls) by computing analysis of molecular variance (AMOVA) in ARLEQUIN package.

3. Results and discussion

A total of 698 distinct Y-STR haplotypes were observed from 938 individuals (Table 1); 588 haplotypes were singletons and 110 haplotypes were shared between 350 unrelated males in Lesotho population as illustrated in Table 1 below.

The most frequent haplotype was shared by 28 unrelated males in Lesotho. The same haplotype was also observed in 17 unrelated males from Bantu population groups in the northern parts of South Africa [1].

A total of nine different duplications were observed at the DYS710 locus (30–33; 30.2–32.2; 30.2–34.2; 31.2–32; 31.2–33 and 31.2–34), six were previously not detected. Three similar duplications were reported in South Africa [4]. All the duplications at DYS710 were associated with a single base deletion at DYS644 giving rise to the .3 microvariants [10]. This observation only occurred in the Southern Sotho (42) and Ndebele (11) ethnic groups. Five individuals with null allele at DYS626 also presented a .3 microvariant at DYS644; however these haplotypes all had a single allele at DYS710. Identical haplotype with a rare duplication at DYS518 (41–42) was also observed among South African Venda [4].

We observed a relatively high level of genetic differentiation amongst the Bantu ethnic groups in Lesotho. AMOVA analysis revealed

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Table 1
Forensic parameters in Lesotho population groups.

	Ethnic groups					
	Southern Sotho	Ndebele	Xhosa	Baphuthi	Vundle	Overall
N	620	120	110	49	39	938
n	481	109	93	42	34	698
DC	0.776	0.908	0.845	0.857	0.872	0.744
RMP	0.03619	0.015417	0.02396694	0.077884	0.061144	0.0337446
HD	0.96536	0.9928571	0.98498749	0.941327	0.963563	0.9671866
FH	22	4	0	2	0	28

N: Sample size, n: number of unique haplotypes, DC: Discrimination capacity, RMP: Random match probability, HD: haplotype diversity, FH: Most frequent haplotype count.

Table 2
: Matrices of Pairwise differences (Rst) (below diagonal) and *Fst P*-values (above diagonal) between Bantu population groups in Lesotho.

	Southern Sotho	Ndebele	Baphuthi	Xhosa	Vundle
Southern Sotho	0.00000	–	+	+	+
Ndebele	−0.00295	0.00000	–	+	+
Baphuthi	0.01859	0.01283	0.00000	+	–
Xhosa	0.08859	0.09046	0.12065	0.00000	–
Vundle	0.10698	0.10442	0.14878	−0.00929	0.00000

(+): significant *P*-values; (-): non-significant *P*-values.

that genetic differentiation was considerably influenced by ethno-linguistic factors (Ethnicity $F_{st} = 0.05343$, $P < 0.001$, and Language $F_{st} = 0.06712$, $P < 0.001$) rather than geographic factors (Districts $F_{st} = 0.00956$ and Regions $F_{st} = 0.01241$, both $P > 0.001$ after Bonferroni correction) (Table 2).

Due to its inheritance mode, Y chromosome haplotypes have a tendency of displaying a population or region-specific clustering effect. In Lesotho, practices such as patrilocal marriages and its clan-based societies especially within the ethnic minorities can be viewed as a major potential contributor to such clustering observations. This can be a major factor affecting DC due to the presence of high frequency haplotypes. The Southern Sotho are the dominating culture and assimilation is expected because of the overwhelming population size differences. However, significant differences with Baphuthi, Xhosa and Vundle do not confirm this expectation.

4. Conclusion

The UniQ Typer™ Y-10 amplification kit provided a higher discrimination capacity in Nguni groups compared to the Southern Sotho population. Population structure in Lesotho populations was highly influenced by culture and language rather than geographic factors. The markers incorporated in the UniQ Typer™ Y-10 also revealed significant ethno-linguistic genetic differentiation among Bantu groups in Lesotho. A significant level of novel variation was detected for loci DYS710 and DYS644. We herein generated the first comprehensive Y-STR population dataset for Lesotho which may be of value for forensic applications and genealogy studies.

Declaration of competing interest

None.

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