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Genetic analysis of autosomal and Y-specific STRs in the Karimojong population from Uganda

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Abstract. In this work, 90 individuals living in Karamoja region were typed for 17 autosomal STRs and 40 males were also typed for 12 Y-STRs. Hardy–Weinberg equilibrium was tested for each autosomal locus and no deviations from equilibrium were observed. For autosomal STRs, our sample shows a combined matching probability of 1 in 6.5×10^{19} individuals and a combined power of exclusion of 0.999999988. Haplotype diversity for Y-STRs was 0.9859 and 32 different haplotypes were detected. Our sample was compared with available autosomal data from sub-Saharan African samples and significant differences were found with Mozambique; Cabinda (Angola); Equatorial Guinea; and with Rwanda. Comparisons with the Y-STR data, revealed large genetic distances between our sample and Mozambique, Cabinda and Equatorial Guinea.

Keywords: STR; Autosomal marker; Y-chromosome specific marker; Uganda; Africa

1. Introduction

The Karimojong are eastern Nilotic pastoral people of NE Uganda. They speak an Eastern Nilotic language of the Nilo-Saharan family. Many years ago, a number of groups of people referred to as the Nilotes migrated from near the Nile valley in southern Sudan

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and Ethiopia toward the south and west. Some of those groups took a south-westerly route, passing through the region that is now Kenya and they ultimately settled on the high, dry plateau which is the Karamoja of today. Nilotes are spread in a region at the fringe of Bantu migration route, and apparently were not touched by the Bantu influence. In this work, we present the first genetic characterization of this population.

2. Material and methods

Blood samples were collected from 90 (40 males) unrelated individuals from Karamoja. DNA was extracted using a standard phenol-chloroform method. Samples were typed using kits: AmpFlSTR Identifiler (Applied Biosystems); Powerplex 16 and PowerPlex® Y Systems (Promega), ran on an ABI310 (Applied Biosystems) and analysed with the Genescan 2.1 Analysis software.

Gene/haplotype diversities and F_{st} or R_{st} genetic distances were calculated using the Arlequin software [1]. In population comparisons, DYS385 was excluded and number of repeats at DYS389I was subtracted from DYS389II.

Table 1
Allele frequencies for autosomal STRs in a sample of 90 Karimojong

CSF1PO		**D2S1338**		**D3S1358**		**D5S818**		**D7S820**		**D8S1179**		**PE**	
7	0.036	15	0.006	14	0.054	8	0.083	7	0.036	9	0.030	5	0.077
8	0.018	16	0.012	15	0.250	9	0.036	8	0.280	11	0.060	7	0.065
9	0.060	17	0.054	16	0.268	10	0.036	9	0.101	12	0.173	8	0.125
10	0.274	18	0.060	17	0.369	11	0.196	10	0.292	13	0.149	9	0.036
11	0.149	19	0.244	18	0.060	12	0.387	11	0.155	14	0.304	10	0.054
12	0.381	20	0.119	**D18S51**		13	0.256	12	0.101	15	0.185	11	0.077
13	0.071	21	0.167	10.2	0.006	14	0.006	13	0.036	16	0.048	12	0.071
14	0.012	22	0.107	12	0.006	**D19S433**		**D21S11**		17	0.042	13	0.083
D13S317		23	0.119	13	0.030	10	0.018	24.3	0.006	18	0.006	14	0.101
8	0.042	24	0.036	14	0.048	11	0.030	25	0.006	19	0.006	15	0.077
9	0.042	25	0.030	15	0.125	11.2	0.012	27	0.060	**FGA**		16	0.060
10	0.054	26	0.036	16	0.149	12	0.107	28	0.208	17	0.006	17	0.083
11	0.286	27	0.006	17	0.202	12.2	0.018	29	0.238	18	0.036	18	0.060
12	0.429	29	0.006	18	0.161	13	0.196	30	0.196	19	0.018	19	0.030
13	0.107	**D16S539**		19	0.119	13.2	0.060	31	0.054	20	0.042	**VWA**	
14	0.042	5	0.006	20	0.077	14	0.339	31.2	0.036	21	0.036	13	0.030
PD		8	0.060	21	0.060	14.2	0.024	32	0.030	22	0.196	14	0.113
2.2	0.220	9	0.149	22	0.018	15	0.143	32.2	0.054	23	0.226	15	0.065
3.2	0.006	10	0.125	**TH01**		15.2	0.036	33.2	0.030	24	0.167	16	0.244
5	0.018	10.1	0.006	6	0.161	16	0.006	34.2	0.018	25	0.042	17	0.250
6	0.048	11	0.268	7	0.482	16.2	0.006	35	0.042	25.2	0.006	18	0.143
7	0.054	12	0.238	8	0.143	17	0.006	36	0.006	26	0.024	19	0.095
8	0.125	13	0.107	9	0.089	**TPO**		37	0.018	27	0.054	20	0.054
9	0.137	14	0.036	9.3	0.089	6	0.048			28	0.071	21	0.006
10	0.101	15	0.006	10	0.036	7	0.006			29	0.036		
11	0.060					8	0.357			30	0.006		
12	0.125					9	0.363			30.2	0.012		
13	0.054					10	0.089			30.3	0.006		
14	0.042					11	0.107			31	0.012		
15	0.012					12	0.030			31.2	0.006		

3. Results and discussion

The allele frequencies obtained for the autosomal markers are those on Table 1. A list of the Y-STR haplotypes in the sample of 40 males can be obtained from the corresponding author upon request.

For autosomal loci, no deviations to Hardy–Weinberg equilibrium were observed. The only *P*-value below 5% was found for CSF1PO ($P=0.0124$) but, if Bonferroni correction is used, the departure is not significant. For autosomal STRs, our sample shows a combined matching probability of 1 in 6.5×10^{19} individuals and a combined power of exclusion of 0.999999988. Y-haplotype diversity was 0.9859 and 32 different haplotypes were detected.

Comparison with available autosomal data from other sub-Saharan African samples showed significant differences with Mozambique [2] in 8 out of 17 loci; Cabinda [3] in 5 out of 17 loci; Equatorial Guinea [4] in 4 out of 17 loci; and with Rwanda [5] in 1 out of 13 loci. Comparisons with the Y-STR data, revealed large genetic distances between the Uganda and Mozambique [6], Cabinda [pers. data] and Equatorial Guinea [7] ($R_{st}=0.168$, $R_{st}=0.195$ and $R_{st}=0.183$, respectively).

The present work confirms the high genetic heterogeneity between African populations and, therefore, emphasizes the importance of using local forensic databases, for both autosomal and Y-specific STRs.

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