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SHORT COMMUNICATION

Allele frequencies of 11 X-chromosomal loci in a population sample from Ghana

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Abstract Eleven X-chromosomal short tandem repeats (STRs) from two multiplex PCR approaches (DXS6807, DXS8378, DXS7132, DXS6800, DXS9898, DXS7424, DXS101, DXS7133, HPRTB, DXS8377, and DXS7423), located in four different X-chromosomal linkage groups, were typed in a population sample from Ghana, Africa. After genotyping unrelated men (129) and women (114) from the Ashanti population, forensic efficiency parameters such as polymorphism information content and mean exclusion chance were calculated. A deviation from the Hardy–Weinberg equilibrium could not be found. The investigation of 11 father–daughter and seven mother–son meioses revealed no mutations in any STR analyzed. Our data were compared with European, African-American, and Asian populations from the literature.

Keywords X chromosome · STR · Multiplex PCR · Population data · Ghana

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Introduction

The analysis of X-chromosomal short tandem repeats (STRs) is especially useful in complex kinship testing, when the offspring is female and the alleged father is missing, or in maternity analyses [19]. In recent years, studies on forensically interesting microsatellite repeats of the human X chromosome have been accumulated [Table S3, 1, 2, 10, 13–16, 20–24]. Nevertheless, results from other than European populations for the majority of X-STRs studied here are still rare and data from larger African populations for these STRs could only be found for an Ethiopian population [7], in one investigation including an Afro-American population [9], and another study from the same working group comprising populations from Angola, Mozambique, and Uganda [10]. Since greater differences in microsatellites and mtDNA between African, Asian, and European populations have been reported (for example [11, 12]), differences in allele frequencies of X-chromosomal STRs could be expected. Here, we present allele frequencies for 11 X-chromosomal STRs from an Ashanti population from Ghana.

Materials and methods

Population

Buccal swabs and blood samples were collected from 129 males and 114 females (randomly selected from a group of 5,100 apparently unrelated adults; 20–48 years old) of the Ashanti population from Ghana. Samples were obtained and analyzed after advice of the Committee on Human Research Publication and Ethics (School of Medical Sciences, Kwame Nkrumah University of Science and Technology, Kumasi, Ghana) and of the Medical Ethics Committees of the University of Duisburg-Essen and University of Schleswig-Holstein in accordance with the declaration of Helsinki. The anonymity of the individuals investigated was preserved corresponding to rules of data protection of the Human Medical Faculties Essen and Kiel. Additionally, 11 girls and seven boys have been analyzed, whose parents were included in the 129 males and 114 females.

DNA extraction

DNA was extracted from buccal swabs with the Invisorb Forensic Kit (Invitec, Berlin, Germany) and the Qiagen Blood Mini Kit (Qiagen, Hilden, Germany). DNA extraction from blood samples was performed with the Qiagen Blood Mini Kit.

DNA amplification and fragment analysis

Two hexaplex amplification reactions were performed as described previously with slight modifications [14]. The first comprised the STRs DXS6807, DXS7132, DXS6800, DXS9898, DXS101, and HPRTB, the second included the STRs DXS8378, DXS7132, DXS7424, DXS7133, and DXS8377. In comparison to the published X-chromosomal amplification assays, we excluded STRs DXS10011—due to amplification problems—and ARA—because of the recommendations of The forensic ChrX research group board as published on http://www.chrx-str.org/. We included STRs DXS6800 (TAMRA-labeled), DXS7132 (TAMRA-labeled), and DXS7423 (FAM-labeled) whose primer sequences have been taken from http://www.chrx-str.org/. DXS7132 was included in both hexaplex amplification reactions (with the same primer sequences) as an internal control against mix-up of samples. PCR fragments were analyzed using an ABI310 Genetic Analyzer (Applied Biosystems, Weiterstadt, Germany) and fragment sizes were determined using the Genemapper ID v3.2 software with especially defined panels and bin sets. The ladder was composed of cell line DNAs and individuals from a German population as described before [14] with the modifications concerning the excluded and included STRs. Allelic nomenclature used for typing was mostly according to Szibor et al. [17] with the exception of DXS9898 in cell line 9948. Here, we found 13 repeats instead of 14 repeats, which is in line with results from Gomes et al. [9]. Additionally, we followed the new nomenclature regarding HPRTB as proposed in three recent publications [Table S3, 1, 9, 13].

Statistical analysis

Allele frequencies, haplotype and gene diversities, exact test of population differentiation and population pairwise genetic distances F_{ST}, Hardy–Weinberg equilibrium in females, analysis of molecular variance, and pairwise exact test of linkage disequilibrium were all performed using the Arlequin ver. 3.0 software [8]. Genetic distance analysis was performed for all X-STRs using F_{ST}, since comparison of F_{ST} and R_{ST} values in Sub-Saharan African populations studied by Caglia et al. [3] showed a better performance for F_{ST} values in an African context. Forensic efficiency parameters were assessed for each locus following Desmarais et al. [4].

Results and discussion

Genotyping of 114 females and 129 males from the Ashanti population of Ghana for the 11 X-chromosomal STRs resulted in allele frequencies and forensic efficiency parameters as shown in Table S1. DXS6807 and DXS6800 were the least informative markers, whereas DXS101 and DXS8377 demonstrated the highest values. Combined forensic efficiency parameters were calculated for all STRs with the exception of DXS7424, for which linkage disequilibrium to DXS101 has previously been detected [5], as follows: combined power of discrimination for males 0.999999, combined power of discrimination for females greater than 0.99999999, combined mean exclusion chance for trios 0.999997, and combined mean exclusion chance in father/daughter pairs 0.9998. The frequency of DXS101–DXS7424 haplotypes is shown in Table S2. Linkage disequilibrium between DXS101–DXS7424 has not been tested for our samples because the number of investigated individuals was too small. The investigation of 11 father–daughter and seven mother–son meioses revealed no mutations in any STR analyzed.

Genetic distances were estimated for each STR marker with the exception of DXS7424—due to the linkage disequilibrium to DXS101—to different European [Table S3, 1, 6, 13, 15, 18], Asian [16], Afro-American [9], and African [10] populations as available (see Table S3 for all results). In general, significant genetic distances were obtained between all European and Asian populations, but

not for the Afro-American population or the three African populations, with few exceptions. In HPRTB, significant genetic distances were found even for Angola, Uganda, and the Afro-American population. For DXS7132, significant genetic distances were scored only for Uganda, and for DXS8378 significant genetic distances were only found for Korea, Latvia, and Germany.

Despite the fact that the X-chromosomal STRs employed in this work have been predominantly studied in European or Asian populations, they proved to be highly discriminating and combined values of forensic efficiency parameters were high in the population sample from Ghana. Therefore, the two hexaplex assays are useful for identification studies involving people from Africa, for example in applications of political asylum in complex deficiency cases.

References

1. Aler M, Sanchez-Diz P, Gomes I, Gisbert M, Carracedo A, Amorim A, Gusma L (2007) Genetic data of 10 X-STRs in a Spanish population sample. Forensic Sci Int 173:193–196
2. Becker D, Rodig H, Augustin C et al (2008) Population genetic evaluation of eight X-chromosomal short tandem repeat loci using Mentype Argus X-8 PCR amplification kit. FSI Genetics 2:69–74
3. Caglia A, Tofanelli S, Coja V et al (2003) A study of Y-chromosome microsatellite variation in sub-Saharan Africa: a comparison between FST and RST genetic distances. Hum Biol 75:313–330
4. Desmarais D, Zhong Y, Chakraborty R, Perreault C, Busque L (1998) Development of a highly polymorphic STR marker for identity testing purposes at the human androgen receptor gene (HUMARA). J Forensic Sci 43:1046–1049
5. Edelmann J, Hering S, Kuhlisch E, Szibor R (2000) Validation of the STR DXS7424 and the linkage situation on the X-chromosome. Forensic Sci Int 125:217–222
6. Edelmann J, Hering S, Michael M et al (2001) 16 X-chromosome STR loci frequency data from a German population. Forensic Sci Int 124:215–218
7. Edelmann J, Lessig R, Hering S, Brundirs N, Kuhlisch E, Szibor R (2004) Allele frequencies for X-chromosomal microsatellites in different populations. In: Doutremépuich C, Morling N (eds) Progress in forensic genetics 10. Elsevier, Amsterdam, pp 263–265
8. Excoffier L, Laval G, Schneider S (2005) Arlequin ver. 3.0: an integrated software package for population genetics data analysis. Evol Bioinform (Online) 1:47–50
9. Gomes I, Prinz M, Pereira R et al (2007) Genetic analysis of three US population groups using an X-chromosomal STR decaplex. Int J Legal Med 121:198–203
10. Gomes I, Alvesa C, Maxzudc K et al (2007) Analysis of 10 X-STRs in three African populations. FSI Genet 1:208–211
11. Herrnstadt C, Elson JL, Fahy E et al (2002) Reduced-median-network analysis of complete mitochondrial DNA coding-region sequences for the major African, Asian, and European haplogroups. Am J Hum Genet 70:1152–1171
12. Kayser M, Brauer S, Schädlich H et al (2003) Y chromosome STR haplotypes and the genetic structure of U.S. populations of African, European, and Hispanic ancestry. Genome Res 13:624–634
13. Pereira R, Gomes I, Amorim A, Gusmao L (2007) Genetic diversity of 10 X chromosome STRs in northern Portugal. Int J Legal Med 121:192–197
14. Poetsch M, Petersmann H, Repenning A, Lignitz E (2005) Development of two pentaplex systems with X-chromosomal STR loci and their allele frequencies in a northeast German population. Forensic Sci Int 155:71–76
15. Poetsch M, Sabule A, Petersmann H, Volksone V, Lignitz E (2006) Population data of 10 X-chromosomal loci in Latvia. Forensic Sci Int 157:206–209
16. Shin HS, Yu JS, Park SW, Min GS, Chung KW (2005) Genetic analysis of 18 X-linked short tandem repeat markers in Korean population. Forensic Sci Int 118:37–40
17. Szibor R, Edelmann J, Hering S et al (2003) Cell line DNA typing in forensic genetics: the necessity of reliable standards. Forensic Sci Int 138:37–43
18. Szibor R, Edelmann J, Zarrabeitia MT, Riancho JA (2003) Sequence structure and population data of the X-linked markers DXS7423 and DXS8377—clarification of conflicting statements published by two working groups. Forensic Sci Int 134:72–73
19. Szibor R, Krawczak M, Hering S, Edelmann J, Kuhlisch E, Krause D (2003) Use of X-linked markers for forensic purposes. Int J Legal Med 117:67–74
20. Toni C, Presciuttini S, Spinetti I, Domenica R (2003) Populations data of four X chromosome markers in Tuscany, and their use in a deficiency paternity case. Forensic Sci Int 137:215–216
21. Turrina S, de Leo D (2003) Population data of three X-chromosomal STRs: DXS7132, DXS7133 and GATA172D05 in North Italy. J Forensic Sci 48:1–2
22. Vauhkonen H, Vauhkonen M, Sipponen P, Sajantila A (2004) Correlation between the allelic distribution of STRs in a Finnish population and phenotypically different gastrointestinal tumours: a study using four X-chromosomal markers (DXS7423, DXS8377, ARA, DXS101). Ann Hum Genet 68:555–562
23. Zalan A, Volgyi A, Jung M, Peterman O, Pamjav H (2007) Hungarian population data of four X-linked markers: DXS8378, DXS7132, HPRTB, and DXS7423. Int J Legal Med 121:74–77
24. Zarrabeitia MT, Alonso A, Zarrabeitia A, Castro A, Fernandez I, Martinez de Pancorbo M (2005) X-linked microsatellites in two northern Spain populations. Forensic Sci Int 145:57–59