ALLELE AND HAPLOTYPE FREQUENCIES OF 6 Y-SPECIFIC STRS (DYS19, DYS389-I, DYS390, DYS391, DYS392 AND DYS393) IN A SAMPLE OF AFRICAN DESCENT INDIVIDUALS OF COLOMBIA

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Y-specific chromosomal STRs have become important in forensic genetics for their potential application in sexual assault cases in order to determine the number of male contributors in a mixed sample, to identify the male DNA profile in a high background of heterologous female DNA and in paternity testing particularly in male lineage cases. In addition, their use in population genetics to trace back the origin of different populations has increased dramatically in recent years. Due to the lack of recombination during meiosis, the "Y" chromosome is haploid in nature and paternally inherited. In order to evaluate the statistical significance of six Y-STRs (DYS19, DYS389-I, DYS390, DYS391, DYS392 Y DYS393), a population database from 123 unrelated individuals of African descent collected in five different towns (Quibdo, the state capital, Condoto, Istmina, Tado, and Bahia Solano) of the Choco department in the Pacific region of Colombia was established.

PCR amplifications were carried out as single loci for DYS19, DYS389-I and DYS392 with identical conditions as described by Kayser et al (1). A multiplex reaction was performed for DYS390, DYS391 and DYS393 (denaturation at 94°C for 3 minutes; 5 cycles of 94°C for 15 seconds, 58 °C for 20 sec., 72 °C for 20 sec., and 30 cycles of 94°C for 15 sec.; 54°C 2º sec.; 72°C 2º sec) and manually analyzed with silver nitrate staining in 4% Acrylamide/bis-acrylamide denaturating gels. Allele designations were made with the aid of sequenced allelic ladders.

Allele and haplotype frequencies (DYS19/S389-I/390/391/392/393), as well as gene diversity were determined. Although the allele distribution was similar to that reported for an African population, some differences were detected.

A total of 80 different haplotypes were found. Fifty-six of them (70%) were single instance haplotypes and 24 (30%) were present in more than one instance. The most frequent haplotype was found at a frequency of 4.87% (6 out of 123) (15/10/21/10/11/14). Two haplotypes were present in five individuals, two haplotypes in four individuals each, five haplotypes in three individuals each one, and fourteen haplotypes in two individuals each one.

Our result shows that the African population studied is genetically heterogenous. The haplotype frequencies obtained will be of great use in anthropological and forensic studies, mainly for sexual assault cases, the identification of human remains and in paternity testing studies.

References

1) Kayser, M., Caglia, A., Corach, D., Fretwell, N., Gehrig, C., Graziosi, G., Heidorn, F., Herrmann, S., Herzog, B., Hidding, M., Honda, K., Jobling, M., Krawczak, M., Leim, K., Meuser, S., Meyer, E., Oestereich, W., Pandya, A., Parson, W., Penacino, G., Perez-Lezaun, A., Piccinini, A., Prinz, M., Schmitt, C., Schneider, PM., Szibor, R., Teifel-Greding, J., Weichold, G., de Knijff, P., and Roewer, L. (1997). Evaluation of Y-chromosomal STRs: a multicenter study. Int. J. Legal Med. 110:125-133.