GENETIC VARIABILITY FOR 25 AUTOSOMAL STR MARKERS, 12 CHROMOSOME X MARKERS AND 30 INDELS IN A BRAZILIAN POPULATION SAMPLE

Mirella Perruccio Soler[1], Carlos Henrique Ares Silveira da Motta[2], Daniela Koshikene[3], Dayse Aparecida da Silva[4], Elizeu Fagundes de Carvalho[4], Gustavo Chemale[5], Hemerson Bertassoni Alves[6], Iraê de Aguiar[7], Jorge M. Freitas[5], Luciano Arantes[8], Laélia Pinto[9], Marcelo Malaghini[6], Renato T. F. Paranaiba[5], Sandra Regina Rachadel Torres[10], Teresa Cristina L. Rocha[7]
[1]QIAGEN Biotecnologia, SP, Brazil
[2]Setor de Genética Molecular - Instituto de Medicina Social e de Criminologia de São Paulo, SP, Brazil
[3]Laboratório de Genética Forense - Polícia Civil do Amazonas, AM, Brazil
[4]Laboratório de Diagnósticos por DNA - Universidade do Estado do Rio de Janeiro, RJ, Brazil
[5]Laboratório de Genética Forense - Instituto Nacional de Criminalística - Diretoria Técnico-Científica - Polícia Federal - Ministério da Justiça - Brasília, DF, Brazil
[6]Laboratório de Genética Molecular Forense - Instituto de Criminalística do Paraná, PR, Brazil
[7]Núcleo de Perícia em DNA Forense do Ceará, CE, Brazil
[8]Instituto de Pesquisa de DNA Forense - Policia Civil do Distrito Federal, DF, Brazil
[9]BIOCOD Biotecnologia, BH, Brazil
[10]Instituto Geral de Perícias de Santa Catarina, SC, Brazil

Introduction: After more than 25 years of hypervariable regions discovery in human genome, human identification by DNA analysis is routinely used as an auxiliary tool for criminal and kinship investigation. At the beginning, there was a limitation on the number of loci commercially available, hindering the achievement of conclusive results. Even nowadays, when polymorphism analysis of chromosome X is needed, there is no commercially available system with known allele's frequency for Brazilian population. Moreover, for over a decade STRs (short tandem repeats) were considered the gold standard for human identification, but in recent years new classes of molecular markers such as SNPs (single nucleotide polymorphisms) and INDELS (insertions/delections) have been presented as options to extend the genomic coverage and overcome some STRs limitations. As an example, a commercial system containing 30 loci INDELS reveals variability in DNA fragments with less than 150 base pairs, helping highly degraded DNA sample analysis.

Objective: Know the allelic diversity and allele frequencies of autosomal, gonosomal and INDEL markers in a Brazilian population sample.

Methods: This study involved 1,000 non related individuals from 10 public or private laboratories covering the 5 Brazilian administrative regions (North, Northeast, Midwest, Southeast and South). DNA was extracted from FTA cards or buccal swabs through FTA purification or Chelex methodology. Kits Investigator Argus X-12 (QIAGEN), Investigator HDplex (QIAGEN), Investigator Hexaplex ESS (QIAGEN) and Investigator DIPplex (QIAGEN) were used according to user's manual recommendations and genotyping of PCR products were made on 310, 3130xl or 3500 Genetic Analyzer instruments (Applied Biosystems). Statistical analysis was performed employing IDproof Software (QIAGEN) and in-house developed software.

Results and Discussion: Brazil has a great admixture population owing to its historical colonization. Microvariants that have been previously described and some new microvariants were detected and will be confirmed by sequencing to determine the cause of variation. Selection of the commercially available kits in this study was due to its lack of population data regarding allele frequencies and its features for forensic applications in Brazil. ⌘