

GENETIC ANALYSIS OF INSERTION-DELETION POLYMORPHISMS FOR FORENSIC USE IN THE URUGUAYAN POPULATION.

André, F.¹, Pagano S.¹, Pisano N.², Sandberg N.¹

¹Dirección Nacional de Policía Técnica – Laboratorio Biológico, Montevideo, Uruguay

²Hospital Italiano - Instituto de Genética Médica, Montevideo, Uruguay

Advancement in forensic population and evolutionary genetic research has been the result of continuous development of new methodologies. The benefits of microsatellites are several and well known. However useful, STRs have some limitations such as the amplicon sizes, which limit analysis of degraded or ancient samples and the “stutter” artifacts which difficult mix stain analysis. Even though SNPs are more suitable for analysis of degraded samples due to the smaller amplicon size and lower mutation rates, this method presents complicated analysis strategies for the detection of different base states. Insertion-Deletion Polymorphisms studies combine the advantages of SNPs, such as short amplicon size, with the added advantage of more discernible size difference between alleles. In addition, IDPs alleles can be detected with the same method as for the STRs typing.

131 unrelated Uruguayan individuals from 4 different geographic areas were assessed for allele frequencies of 30 insertion-deletion polymorphism markers previously selected and validated for forensic purposes (Investigator DIPplex kit, QIAGEN). Said markers are distributed over 19 autosomes that are at least 10 Mbp away from any commercially available STR and SNP marker. The genetic structure of the populations and basic parameters of molecular diversity, including analyses of molecular variance (AMOVA) were studied. No significant difference was detected among southern, littoral, central and northern populations. Hardy-Weinberg equilibrium tests and linkage disequilibrium analysis were performed and the allele frequency distributions of the 30 InDel markers met the conditions for genetic equilibrium and the InDel markers on the same chromosome did not generate any linkage blocking. The combined power of discrimination was 0.99.

Also, results of real cases degraded sample analysis using STR markers were compared with those obtained with InDel analysis in order to validate performance on compromised samples. Results show that InDel analysis yields more genetic information from complex cases such as ancient bones and low concentration samples and provide information to complement those results obtained by STR analysis in complex relationships cases as well. ☒