THE HUMAN Y CHROMOSOME: DISTRIBUTION OF 16 MICROSATELLITE MARKERS IN THE PORTUGUESE POPULATION

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In this work we demonstrate the usefulness and informative power of Y-STRs in the Portuguese male lineage identification. For this aim a total of 658 individuals from the Portuguese population were typed for the loci: DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4 and GATA H4. The analyzed samples included males from all 18 districts that subdivide Portugal. Of the typed population, 316 individuals belonged to the north region, that includes districts located above the Douro River, 230 from the center, with districts situated between Douro and Tejo Rivers and 112 came from the south part of the country. Genetic distances between the 18 districts studied were estimated. Results obtained by this analysis revealed the proximity between populations, except for the Portalegre district with significant Rst ($P<0.05$) values, varying between 0.06723 and 0.1467. Analysis of molecular variance (AMOVA) was used to evaluate male population substructure of Y-STRs haplotypes. Results support the homogeneity of the Portuguese population and most of the molecular variation was due to variation within districts (99.29%) rather than among them. Haplotype and allele frequencies of these sixteen Y-STRs were estimated in all 3 regions. The lowest haplotype diversity was observed in the south region of Portugal (0.9992) followed by the center (0.9995) and finally by the highest value in the north region of the country (0.9996). Despite of a small increase of the haplotype diversity in the north region, haplotype analysis of these markers showed the relative homogeneity of the country with an overall haplotype diversity of 0.9997. The analysis of this population of unrelated males sampled from the 3 regions of Portugal revealed a total of 602 different haplotypes. According to the results in a sub-sample of 479 individuals typed in a previous study for the genetic markers included in the "Y-STR Haplotype Reference Database" (http://ystr.org, http://ystr.org/usa e http://ystr.org/asia) (Gusmão et al., VII Jornadas de Genética Forense, Barcelona 4-7/06. Abstract P46) and those observed in this work, the inexistence of substructure of the Y-STR haplotypes between (and inside) these three regions is compatible with the use of a single database in Portugal for forensic purposes.