

Worldwide diversity of the Y-chromosome tetra-local microsatellite *DYS464*

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ABSTRACT. Of all DNA markers on the human Y-chromosome, the tetra-local Y-linked microsatellite *DYS464* is the most polymorphic. We genotyped *DYS464* in 677 male samples collected worldwide, maintained in the HGDP-CEPH Human Genome Diversity Cell Line Panel. Fourteen different alleles were found, with allele lengths varying from 9 to 23 repeats. One hundred and seventy-five different genotypes were detected, of which 90 appeared to be continent-specific. The region with the highest percentage of unique genotypes was Africa. Genotype diversity was 0.98 for Europe, 0.97 for Central and East Asia, 0.95 for Africa, 0.94 for Oceania, 0.92 for the Middle East, and 0.90 for the Americas. A hierarchical analysis of molecular variance showed low levels of worldwide genetic structure; 88.42% of the genetic variance was found within populations, 9.62% between populations within regions and 1.96% between regions. Since the four *DYS464* repeats are identical, one cannot assign each peak in the electropherogram to a specific locus. Thus, the same genotype may correspond to several haplotypes, with different permutations of alleles. Consequently, genotypes are degenerate, which limits phylogeographical analyses. Yet, because of its high variability, *DYS464* still constitutes an informative tool for population and evolutionary studies.

Key words: Y-chromosome; Worldwide populations; Microsatellites; DNA; Population genetics; *DYS464*