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The use of the Y chromosome in forensic genetics—current practices and future perspectives

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There are a number of merits that qualify the Y chromosome as a special forensic genetic tool: the male specificity for most of its length, the absence of recombination which provides unambiguous male lineages and the small effective population size that tends to create population-specific allele distributions on the Y chromosomes.

Since the main goal of forensic genetics is individualization of persons or lineages of descent, an analytical strategy for the male chromosome must enable the expert to differentiate between the majority of unrelated haplotypes. For this to achieve the choice of the sequence type and its variability (i.e. its mutation rate) as well as the number of individual sequences to be used for profiling is crucial. We have introduced a short tandem repeat (STR) profile for the Y-chromosome consisting of 11 microsatellite sequences, which is both informative for individualization purposes as well as for a genetic distance analysis of populations. The technical simplicity of the approach led to a rapid introduction of the technique in many of the forensic labs worldwide. Intense international collaboration facilitates the generation of large haplotype reference databases, most of them are available online and searchable (Europe: http://ystr.org/europe and Asia: http://www.ystr.org/asia/). By use of haplotype-specific parameters such as the molecular distance (which equals the minimum number of mutational steps separating two haplotypes) and the largest available haplotype databases, a Bayesian approach to evaluate Y-STR haplotype matches has been proposed [1,2].

The recommendations of the ISFG [3a,b] state some basic principles on forensic analysis using Y-STR polymorphisms: the use of sequenced allelic ladders (now commercially available), the application of a repeat-based nomenclature and the use of suitable haplotype reference databases for statistical evaluation of matches.

Still a matter of research but of the utmost interest is the potential of the Y-chromosome analysis to unravel the ethnological background of a given male profile. A dual

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approach—that using Y-STRs as well as Y-SNPs—probably renders the maximum amount of information about the descent of a male lineage typed in a forensic specimen [4].

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