Y-SNPS AND FORENSIC APPLICATIONS: A PRELIMINARY STUDY

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Low mutation rate, paternal heritage only and absence of recombination, make Y-SNPs particularly suitable for evolutionary studies. Moreover, many of them show regional specificity, providing useful information about the geographic origin of a subject or evidence under investigation. The microsatellites of the Y-chromosome are useful in forensic casework analysis involving male subjects, such as paternity testing, identification of bloodstains or other biological evidence with mixed profiles and reconstruction of family relationship by patrilineage analysis.

Such biallelic polymorphisms are of special interest for forensic purposes, because the amplification of short fragments including the single base mutation which characterizes their polymorphism potentially allows a positive result even with high-degraded DNA, where microsatellites fail. In casework, where decomposed bodies or human remains, or victims of mass disasters, including aircraft or boat accidents involving people from various geographical areas, are analyzed, the detection of geographical origin is a powerful aid to forensic investigation.

The aim of this study is to set-up multiplex PCR of NRY SNPs suitable for forensic purposes and evolutionary studies. A first multiplex with 7 SNPs (M35, M89, M172, M170, M9, M173 and M45) which occur in the basal branches of the phylogenetic tree, and able to assign a subject to known European haplogroups was designed.
SNP genotyping was accomplished by hot-start PCR with primers amplifying fragments between 96 and 136 nucleotides, minisequencing, and capillary electrophoresis of extension products.

Experiments on low molecular weight DNA were performed on DNA extracted from whole blood at a concentration of 20 ng/μl and fragmented in a sonicator. Sonicated DNA was submitted to minisequencing and compare to DNA amplified with the AmpFlSTR® Identifiler™ PCR Amplification kit (Applied Biosystems).