PYROSEQUENCING™ TECHNOLOGY FOR FAST AND DEPENDABLE DETERMINATION OF DNA SEQUENCE VARIATION

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Forensic applications have much to gain from an efficient exploitation of the DNA sequence variability provided by single-nucleotide polymorphisms (SNPs) and insertion/deletion polymorphisms. As an example, investigations of mitochondrial DNA have pinpointed highly variable regions that maximize the discriminatory power obtained per determined DNA base. The implementation of new techniques, dedicated to SNP and short sequence determination, may revolutionize forensic testing by enabling delivery of these types of data within a few hours of time. Pyrosequencing™ technology provides a sequencing-by-synthesis procedure that is highly automated and optimal for analysis of short to medium length DNA sequences. Through its real-time base-calling process, Pyrosequencing™ technology combines the discriminatory ability of conventional DNA sequencing with the speed of recently developed techniques. The method is being widely applied in the contexts of signature tag determination and SNP genotyping in haploid or diploid genomes, and has proven suitable for routine forensic analysis of mtDNA samples. Its quantitative output ensures straightforward assessment of heterogeneity within samples, such as heterozygous genotypes, heteroplasmia, or impurity of samples. We present the principles of Pyrosequencing™ technology and outline the areas within forensic science where the method is likely to be at a particular advantage.