

# Somatic and Germline Mutation of Forensic DNA Markers

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**REFERENCE:** Yang C-H, Staropoli T: Somatic and germline mutation of forensic DNA markers; *Forensic Sci Rev* 13:131–152; 2001.

**ABSTRACT:** Somatic and germline mutations of forensic DNA markers DQA1, PM, STR, minisatellite DNA, and mtDNA are reviewed in this paper. DQA1 and PM are stable point substitution polymorphic loci in nuclear DNA (nucDNA); thus, somatic and germline mutations are rarely reported. STR loci somatic mutations are not found in normal cells but are very unstable in cancer cells. Germline mutations are detected in STR loci with a mutation rate of approximately 0.1%. Germline mutations of STR loci are nucleotide sequence, sex, and age dependent. Among the 13 CODIS core, TH01, TPOX, D16S539, and D3S1358 are more stable than others. Both somatic and germline mutations are found in minisatellite loci. Somatic mutations at mtDNA central region (CR) are detected in different tissues within the same individual, and the mutation rates are high in several cancer tissues. The intergenerational mutation rate of mtDNA CR is estimated to be 1/400 to 1/1200 per generation by phylogenetic analysis. The intergenerational mutation rate estimated by the recently developed pedigree method is higher, 14/1221 (1/87). This rate does not include the hot spot and heteroplasmic mutation. If substitution, heteroplasmic, and poly-C tract mutation are included, the mutation rate of mtDNA CR is 33/1590 (1/49) per generation. Proposals and practices on the interpretation of test data that may be associated with mutation are also discussed.

**KEY WORDS:** DQA1, Germline mutation, minisatellite DNA, mitochondrial DNA (mtDNA), polymarkers (PM), short tandem repeat (STR), somatic mutation, variable number tandem repeat (VNTR).

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