# Disease-Induced Anomalous Human Microsatellite DNA — Implications in Forensic DNA Typing

**REFERENCE:** Pai C-Y: Disease-induced anomalous human microsatellite DNA — Implications in forensic DNA typing; *Forensic Sci Rev*; 18:43–58; 2006.

**ABSTRACT:** Short tandem repeat (STR) markers adopted for forensic applications are generally stable and have been proved effective for the analysis of many archival pathology specimens and postmortem tissues. However, recent studies on STR typing of cancerous samples have shown misinterpreted profiles caused by two types of genetic alterations in tumor materials: microsatellite instability (MSI, contractions or expansions of a heterozygous allele) and loss of heterozygosity (LOH). Forensic STR typing of tumor material is unlikely; however, under rare circumstances, these may be the only samples available. This article reviews literature information in the following areas: unusual sample types encountered in casework; possible causes of STR alterations; genetic diseases associated with CODIS 13 markers; and forensic evaluation of commercial STR markers used in solid tumor tissues. Literature information suggested that STR-associated events observed in unusual cases may not be readily explained by known biological and disease processes. In actual practice, false profiles would only arise from complete LOH (rather than allelic imbalance) and MSI. Adoption of different analysis methods and various sets of genetic markers in profiling may lead to inconsistent results. When comparing profiling data reported by different laboratories, one should also note whether the same methodology was applied to the same set of genetic markers used for the evaluation of the disease tissues. To avoid misinterpretations, each tumor type should be evaluated individually, as various disease types may exhibit distinct behavior nature.

**KEY WORDS:** Anomalous DNA profile, cancer, CODIS, disease, forensic STR marker, loss of heterozygosity (LOH), microsatellite instability (MSI).