LOSS OF HETEROZYGOSITY DETECTED IN A SHORT TANDEM REPEAT (STR) LOCUS COMMONLY USED FOR HUMAN DNA IDENTIFICATION

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Short tandem repeats (STR) testing is the "gold standard" for human identification testing. STRs can also be used to study genetic instability of chromosomal loci. A genetic deletion common to many types of cancer is referred to as the loss of heterozygosity (LOH). Numerous examples of LOH in cancer have been described and some have been mapped to areas located in close proximity to markers employed in human identity testing. LOH has rarely been observed for STR loci commonly employed in forensic testing. Recently, for medico-legal purposes, we were asked to determine whether a tissue biopsy originated from a particular individual. For a reference source we assessed two specimens, one from normal tissue and one from cancerous tissue. When both reference specimens were used to generate DNA profiles we observed LOH at one STR locus, D13S317. As demonstrated in other cancers, only the cancerous biopsy demonstrated LOH. The forensic community should be cognizant of these unusual circumstances because, as identification of human DNA continues to be used more extensively, certain instances will arise in which reference material will not be readily available. In these situations, archived specimens may be employed as a reference source. Clinical specimens such as tissue biopsies should be used with caution if they have not been confirmed to contain normal tissue.