LOW COPY NUMBER DNA ANALYSIS: PROCEDURE AND PRACTICE

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The phenomenal success of DNA profiling has increasingly led to attempts to analyze more difficult and challenging samples to include samples containing DNA from only a few cells. Attempts to increase the sensitivity of the STR assays by increasing the number of PCR cycles were first described more than a decade ago. This approach, which has become known as low copy number (LCN) DNA typing, has led to the analysis of a new category of samples called variously 'touch', low-template or high sensitivity.

The limitations of LCN arise from the exaggerated stochastic effects seen with the very low levels of input template DNA. Concerns have been raised that the laboratories formally conducting LCN analyses perform casework in a manner that is inconsistent with their protocols and/or is not supported by validation studies. A case example from New Zealand will be described. The ESR laboratory reported YSTR results of two facial cheek swabs from a deceased woman, each with only one locus showing a peak. The two total 'allelic' peaks, observed at two different loci, were seen in only ***one*** of four replicates. These peaks had heights of 54rfu and 70rfu and no other peaks were detected at any of the other loci. The possibility of contamination was not discussed in the report and the single non-replicating peaks were deemed alleles. According to recommendations requiring replicate alleles these peaks should not have been reported. Additionally this information was used to exclude one man and include a second man as the source of the DNA on the cheek swabs. The included man had only one of the two 'alleles' detected.