

PEAK HEIGHT DISCORDANCE AT THE D8S1179 LOCUS – IMPLICATIONS IN THE INTERPRETATION OF DNA PROFILES

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In crime scene samples from 3 recently processed cases, heterozygous peak height concordance values of less than 33% were observed at the D8S1179 locus using the AmpFISTR® Profiler Plus™ system. There was no evidence of a mixture at any other locus in any of the samples. The purpose of this poster is to present the profiles in which severe discordance was observed and to present the rationale underlying their interpretation.

Forensic laboratories must develop guidelines based on validation studies, including criteria for identifying mixtures, to facilitate the interpretation of DNA results. In addition to the presence of additional alleles, peak height concordance within a locus is a major indicator of mixtures. The STR Interpretation and Reporting Guidelines from the Centre of Forensic Sciences, with respect to an evaluation of whether a sample is from a single source, read as follows:

A single source DNA profile will not (barring extremely rare genetic anomalies) exhibit more than 2 alleles at any one locus, and will usually exhibit peak height concordance of >60% within heterozygous loci. The determination of a profile as having originated from a single source will be made having considered these factors in the profile as a whole. For instance, discordance at a single heterozygous locus is not in and of itself sufficient grounds to declare a mixture, when the remainder of the profile meets the criteria expected of a single source sample. Also, the presence of a three-banded pattern at a single locus, given no other indication of a mixture whatsoever, would be indicative of a genetic anomaly at one locus as opposed to a mixture.

While guidelines are necessary, they cannot replace the expertise that a trained and experienced analyst brings to bear on issues of interpretation. A strict reliance on guidelines as “rules” as opposed to guiding principles may have resulted in the above-noted profiles (below the 60% concordance threshold at D8S1179) being falsely interpreted as mixtures.

Each sample was re-amplified using the Promega PowerPlex®16 kit. In this system, all samples showed expected peak height concordance results, confirming that primer-site mutations, specific to the D8S1179 primers in the AmpFISTR® Profiler Plus™ kit resulted in the atypical profiles.