



G29 An Atypical STR Genotype, Including a Three-Banded Allelic Pattern, From a Biopsy Tissue Sample

Sobia Malik, MSc, Centre of Forensic Sciences, 25 Grosvenor Street, 4th Floor, Toronto, Ontario, Canada*

The goal of this paper is to present to the forensic community an instance of a STR genotype that may have been misinterpreted as a mixture. The profile was confirmed by a comparison sample and included allelic peak height discordance and a discordant three-banded allelic pattern using the AmpFISTR ProfilerPlus™ system.

In support of a medical diagnosis carried out at a local hospital, the Centre of Forensic Sciences (CFS) was asked to identify the source of two biopsy samples due to a potential mix-up of samples using a third biopsy sample from a known individual. The results resolved the mixup and in the process also identified, in a single source sample, allelic peak height discordance at amelogenin and vWA as well as a discordant three-banded allele pattern at D18S51.

Two paraffin blocks were submitted. The first contained two biopsies, one malignant and one benign, which raised suspicion that the two biopsies were from different individuals. The second block contained a malignant biopsy sample from a known source. The three biopsy samples were extracted using a xylene extraction technique followed by amplification of 1ng of DNA in the AmpFISTR Profiler Plus™ system. The paraffin block with the malignant and benign biopsies showed two different DNA profiles confirming that these biopsies were from different people.

One of these profiles showed peak height concordance of only 19% at amelogenin and 32% at vWA, as well as a 25% peak height concordance between two alleles as compared to the third allele in a threebanded allelic pattern at D18S51. Based on both internal and published validation studies, the presence of such anomalies is atypical when dealing with a sample known to have originated from a single individual. The fact that the anomalies are compounded is even more rare. CFS internal validation of the STR loci used in the AmpFISTR Profiler Plus™ system has shown that minimum peak height concordance observed at most STR loci where 1ng of single-source DNA has been amplified is 60%. Interpretation of this sample as an unknown or questioned sample would indicate a mixture. However, the comparison sample (of known origin) in the second block also showed peak height discordance at similar percentages at amelogenin, vWA and at the discordant three-banded allelic pattern at D18S51. The peak height discordance at amelogenin, vWA and discordant three-banded allelic pattern at D18S51 may indicate a chimeric genotype since the anomalies are seen at more than one locus and therefore occur at more than one chromosome. It is also possible that more than one genetic event may account for the observed profile.

This example emphasizes that caution should be taken when using tissue block samples as comparison samples in forensic casework. Information that a tissue sample is cancerous may not always be available. Therefore, when forensic DNA analysis involves tissues (a notable example is mass disaster identification), issues involving identification, interpretation and statistical significance regarding atypical profiles and three-banded allelic patterns may become increasingly important.

Biopsy, Three-Banded Pattern, STR