MULTIPLE ALLELES AT FOUR STR LOCI: CONTAMINATION OR...?

Barbara J. McCue, M.T., Ronald J. Rubocki, Ph.D., Shirley J. Shepherd, M.S., James L. Wisecarver, M.D. Ph.D.
Human DNA Identification Laboratory, Department of Pathology and Microbiology, University of Nebraska Medical Center

Analysis of multiple loci using sort tandem repeats (STR) is valuable in human identity testing because a high degree of discrimination among individuals may be obtained. We use the AmpF/STR Profiler™ PCR Amplification Kit (PE-Applied Biosystems) which consists of a fluorescently-labeled multiplex of nine loci in addition to the gender marker, amelogenin. The detection method used in our laboratory is capillary electrophoresis (CE) on an ABI™ Prism® 310 Genetic Analyzer. We received a criminal forensic case which included several pieces of evidence and reference blood samples. After initial testing, one of the suspects had a DNA profile which included 3 alleles at four of the nine loci tested (vWA, FGA, TH01, D5S818). At each locus, two of the alleles appeared to be "major" alleles with a third "minor" allele present. It appeared to be a mixture of two people. The original specimen from which DNA was extracted was a piece of gauze on which a sample from an EDTA tube had been spotted and dried at another laboratory. Contamination of the sample was suspected and we requested a second, unopened blood specimen from this individual. The DNA profile from this second specimen was identical to that of the original specimen. One piece of evidence, a scraping taken from under the suspect's fingernails which was positive for hemoglobin, was also analyzed and the DNA profile matched that of the two reference specimens mentioned above. The relative peak heights of the two "major" and one "minor" alleles remained constant in all three samples. Additional background information revealed that the suspect had not received a bone marrow transplant or blood transfusion. However, we discovered that the suspect is a fraternal (dizygotic) twin. We hypothesize that an exchange of blood cells between the fetuses occurred in utero and that the third "minor" allele in the first twin's reference sample may be an allele that originated from the second twin. No additional specimens from the suspect or his twin could be obtained without a court order. Several important questions remain unanswered. Forensic scientists should be aware of this possible explanation when faced with DNA profile in which extra alleles at multiple loci are detected.