RESOLUTION OF A TYPING DIFFERENCE BETWEEN PERKIN - ELMER'S AMPF/STR™ PROFILER KIT AND PROMEGA'S POWERPLEX™ 1.1 KIT USING SEQUENCE ANALYSIS

David Parsons, BS, Bill Watson, MS, Robert Giles, Ph.D., Du Burt, Ph.D.
GeneScreen

Two related paternity samples were typed using Perkin-Elmer's AmpF/STR™ Profiler Plus™ Kit, resulting in a homozygous single exclusion at vWA. Additional testing with Promega's PowerPlex™ 1.1 Kit produced heterozygous typings with a shared allele at this same locus. These observations indicated a probable null allele with the Profiler Plus™ Kit that typed as the shared allele with the PowerPlex™ 1.1 Kit. Sequencing experiments were undertaken to examine the flanking regions of the vWA locus of both samples for the presence of a mutation that would account for the null allele.

For sequencing, primers were selected so as to generate an amplified product that would include the priming regions for both kits. After amplification, the alleles were separated using electrophoresis and electroelution. Each allele was sequenced and variation from the published vWA sequence was observed in the shared allele. Since the sequences of the primers for the Profiler Plus™ Kit are not published, the exact location of the mutation with respect to the primer cannot be known. If this mutation occurred in the priming region for Perkin-Elmer's Profiler Plus™ Kit, it would provide the needed explanation for the observed results.