B143 Development of a Haplotype Database for the AmpF*l*STR® Yfiler™ PCR Amplification Kit

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Attendees will receive an overview of the development of a haplotype population database for the AmpF*l*STR® Yfiler™ PCR Ampliification Kit and the web-based tool for searching the haplotype database.

This presentation will impact the forensic community and/or humanity by informing the forensic community of a new tool for the analysis of haplotypes obtained using the Yfiler™ PCR Amplification Kit.

This presentation will discuss the development of a haplotype population database for the AmpF*l*STR® Yfiler™ PCR Amplification kit. Attendees of the presentation will be provided with an overview of a webbased tool for searching the haplotype database. Statistical analyses of the population database and the detection of microvariants will be discussed.

Analysis of sexual assault evidence using currently available autosomal amplification and typing systems is complicated by the inability to obtain male DNA profiles in the context of high quantities of female DNA. As a result, sexual assault evidence containing low numbers of spermatozoa or semen from azoospermic or vasectomized males may not be analyzed and, if analyzed, often does not yield male DNA profiles. Analysis of male-specific DNA markers can provide evidence profiles that may have probative or exculpatory value in such cases. Y STR analysis has proven to be a valuable tool for recovering information from evidence samples containing marginal amounts of male DNA. Due to the genetically linked structure of Y STR haplotypes, estimation of population frequencies for a particular haplotype requires analysis of population database samples using the complete set of markers used to analyze the forensic evidence sample. The AmpF*l*STR□ Yfiler™ PCR Amplification kit may also be utilized for paternity analysis in situations involving male offspring, providing an additional tool for discrimination that will complement autosomal STR analysis. The AmpF*l*STR□ Yfiler™ PCR Amplification kit features 17 male-specific DNA markers located on the Y chromosome. A small range of amplicon sizes, from 100-326 base pairs, has been achieved by labeling the amplified products with four fluorescent dyes, including 6-FAM™, VIC□, NED™, and PET□. The selected markers include the European minimal haplotype consisting of DYS19, DYS389I/II, DYS390, DYS391, DY6S392, DYS393 and DYS385a/b, the additional SWGDAM recommended loci, DYS438 and DYS439, as well as six additional loci, DYS437, DYS448, DYS456, DYS458, DYS635 (Y GATA C4), and Y GATA H4. The added loci significantly increase the discrimination potential of the Y haplotype. A haplotype database was developed in order to provide a useful tool for estimating Y haplotype frequencies in a variety of ethnic groups.

Population samples from North American, European and Asian population groups were analyzed by various test site laboratories and compiled on the basis of ethnic group. The data are accessible via the Internet for haplotype searches. Features of the search tool include the ability to input evidence profiles both singly and in batches via automatic upload from Genotyper® software files. The tool is also compatible with data generated using GeneMapper® *ID* v3.2 software. A manual input mode allows entry of microvariant data resulting in the capability for all profiles to be searched and reported. Search outputs provide estimated frequencies for individual ethnic groups as well as for all population samples. Any haplotype matches to the forensic unknown will also be shown. As is the case for mitochondrial DNA, haplotype frequencies are estimated by counting the number of times the haplotype is seen in various populations. Therefore it is desirable to increase the size of the haplotype database where possible. Periodic updates to the database will reflect the ongoing analysis of additional population samples.

Y STRs, Haplotype, Database

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