B9 A Comparative Study of Y-STR Loci: How do Different Sets of Y-STRs Fare on a Common Population Panel?

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Via this presentation, attendees will be presented with the results of a comparative analysis of several sets of Y-STR loci. A new set of YSTR loci developed using the human genome sequence is compared with the most widely used Y-STR loci and with other recently identified Y-chromosome loci.

A number of Y-STR loci have been identified and characterized; however, there are concerns associated with some loci. We address these concerns, identify new Y-STR loci, and perform a side-by-side examination of variability of new Y-STR loci with the most widely used YSTR markers and with a panel of other recently identified Y-STR loci. Since it is important to identify which loci are the most appropriate and informative for forensic applications, this study should have an impact on the forensic community.

Proposition: Additional Y-chromosome microsatellite loci may be needed for adequate forensic analysis for the following reasons: First, the chromosomal distribution of currently available loci is mostly limited to two small regions on the Y-chromosome; second, additional loci may be needed because of potential typing errors caused by duplicated genetic material in the human genome; third, the loci may be needed, as well, because of the relatively low level of variability of the currently available unilocal loci.

Sex specific markers, such as Y-STRs, are highly valuable tools in DNA forensics because men commit the majority of violent crimes. Y-STRs can be used to distinguish the male component in body fluid mixtures. They also aid in the identification of the number of male contributors in multiple rape cases. Y-STRs are also useful in paternity cases; particularly in situations involving a deceased putative father, Y-STRs can identify patrilineage. In population studies, Y-STRs help to identify paternal migration patterns, in contrast to maternal migration patterns identified by mitochondrial DNA.

Six studies involving the identification/characterization of forensically useful Y-STRS are relevant: Kayser et al., 1997, White et al., 1999, Ayub et al., 2000, Iida et al., 2001 and 2002, and Redd et al., 2002. In terms of physical location within the Y-chromosome, loci in the first five studies are mostly limited to two small regions fairly close to the Y-chromosome centromere. Several loci identified by Redd et al., 2002, are also located in these same regions of concentrated loci. In addition to the distribution within the Y-chromosome, a number of loci have additional drawbacks. Examination of information from the human genome project and from the literature indicates that many current loci are duplicated elsewhere on the Y-chromosome or on the X-chromosome. Some of these loci may be highly variable but, for forensics, are less than ideal. Potential problems can occur during interpretation of genotype results. For many forensic applications, the purpose of using Y-STR loci is to preferentially amplify the male DNA contribution in mixed samples and to determine the number of male contributors in multiple rape cases. Duplicated loci defeat these purposes.

Following the identification of loci by the first three studies, various combinations of markers have been used to examine populations, revealing a high number of unique haplotypes within populations. The most widely-used Y-STRs are those of Kayser et al. Given potential drawbacks of some available loci, we have identified new Y-STR loci. About 26 Mb of Y-chromosome DNA sequence has been annotated by the National Center for Biotechnology Information (NCBI). We screened over 17 million bases of Y-chromosome sequence outside the two concentrated regions of existing loci, identifying a library of 465 loci. BLAST searches against the human genome identified those that were unique to one Y-chromosome location. Of 229 loci examined, 63 were determined to be unique to single Y-chromosome locations. The 63 loci were screened in a racially diverse panel of 30 individuals to ensure unique amplification and to determine locus variability levels. Nearly half were found to be highly polymorphic. The loci were further tested for non-amplification in females. As a result, we chose a set of 10 male specific marker loci dispersed across the Y-chromosome. We found 30 unique haplotypes in the 30 individual test population. Two Y-plex kits exist which, together, contain the most frequently used loci. We examined the same 30 individuals with these kits. Several haplotypes occurred multiple times. The OSU 10-locus set had an average of 3 more alleles per locus than the 10-locus Y-plex set. Since we identified over 60 loci by screening the human genome sequence, we expected to find loci in common with other researchers. Examination of the loci identified by Redd et al. 2002, show 7 loci in common between our 63 loci and the 14 Redd et al. loci. The OSU 10-locus set has an average of 2.5 more alleles per locus than the Redd et al. 7-locus set. (Note that only one locus is common to both the Redd et al. 7-locus set and the OSU 10locus set). Further comparative analyses of the different sets of loci will be presented.

A number of Y-STR loci have been identified and characterized; however, there are concerns associated with some loci. We address these concerns, identify new Y-STR loci, and perform a side-by-side examination of variability of new Y-STR loci with the most widely-used YSTR markers and with a panel of other recently identified Y-STR loci. Since it is important to identify which loci are the most appropriate and informative for forensic applications, this study should have an impact on forensic science.

 ** Presenting Author*

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