



B62 Canvassing the Y-Chromosome: The Identification of Genetically Diverse Y-STR Loci

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The goals of this presentation are to present to the forensic community the identification of new Y-chromosome short tandem repeat (Y-STR) loci with forensic potential. These new loci are distributed over the length of the Y-chromosome. Polymerase chain reaction (PCR) amplification for these new loci occurs exclusively in male individuals without the production of products from related loci elsewhere in the genome. Multiple products can occur as a result of genetic duplication events (on the Y-, Xor autosomal chromosomes) during human genome evolution.

Additional Y-chromosome microsatellite loci may be needed for forensic analysis because of the chromosomal distribution of currently available loci and because of potential typing error caused by the occurrence of duplicated genetic material in the human genome.

Men commit a substantial majority of violent crimes. Therefore, Y-STR loci are valuable tools in forensic science. They are particularly useful in the identification of the male contribution in mixed forensic samples. This is due to the fact that the differential lysis step, in which a limited and/or degraded male component has the potential to be lost, is not necessary for Y-STR analysis. In multiple rape cases, Y-STR loci would aid in the identification of the number of perpetrators. In addition to the benefits associated with the analysis of violent crimes, Y-STRs can aid in the identification of paternity, particularly for cases in which the putative father is deceased, to identify patrilineage. They are also useful for population studies. Paternal migration patterns can be analyzed, as well as relationships within and between populations.

As of August 2002, a number of Y-chromosome STR loci have been identified and examined in several populations. Most analyses were performed using different combinations of the loci characterized by Kayser et al., in 1997. The studies revealed a high number of unique haplotypes. Over the last 3 years, several investigators have described additional loci. A few of the loci identified in 1999 and 2000 were found to be informative, particularly when combined with the most informative of the original set of loci. More recently, two of the loci identified in 2002 were included in a 20-plex system with several loci from previous studies.

There are potential problems with these loci, however. Though several loci are informative, they are located mostly within 2 small regions of the Y-chromosome. The Y-chromosome consists of ~59 million base pairs. The currently available loci make up less than 10% of the chromosome. In addition, several of the existing loci are duplicated on the Xand/or Y-chromosome. In fact, several homologous regions exist between the Xand Ychromosome, which may have the potential to undergo recombination. Since the current Y-STRs are not dispersed randomly across the Y-chromosome, if recombination is occurring it will go undetected. The utilization of Y-STRs to identify male individuals is a novel approach. However, the use of duplicated loci in forensic cases is less than ideal, particularly during the identification of the number of perpetrators in a multiple rape case.

A large portion of the Y-chromosome remains to be studied. More than 17 million nucleotides of the Y-chromosome, which were annotated and released in the public database, GenBank, have been screened. Outside of the regions that contain the existing loci 465 were identified. These new loci were evaluated by comparison against the draft version of the entire Human Genome Sequence. Of 229 loci examined to date, 72% are duplicated elsewhere in the human genome, mostly on the Xand/or Y-chromosome. Due to the fact that the present version of the Human Genome Sequence is a draft version, the remaining 64 unique trito hexanucleotide repeat loci were further analyzed in a racially diverse population of males and females to ensure that amplification occurs at only 1 locus and is limited to male individuals.

The loci were also examined in this sample population to assess their forensic potential. The majority of the loci identified possess more than one allele. As of August 2002, several loci which possess 9 alleles in a small test population of ~30 male individuals have been identified. Results will be discussed concerning the genetic diversity of the loci. The most informative loci will be multiplexed and further examined in a larger population of racially diverse male and female individuals.

Y-Chromosome, Y-STRs, Microsatellites