POLYMORPHISM OF NINE STR LOCI ON X CHROMOSOME IN KOREANS

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Compared to autosome or Y chromosome, the number of X chromosome markers is limited. The usefulness of the STR loci on the X chromosome in forensics seems to be restricted, but it may be valuable in several specific situations, for example to confirm either grandmother-grandchild. As males are hemizygous for the X chromosome, the MEC (mean exclusion chance) may be higher than diploid autosomal STRs. The X chromosomal STRs can also be used when the disputed child is a girl. In this polymorphism on nine STR loci in Koreans located on the X chromosome were reported.

Nine STR loci, DXS6803, DXS8378, CHLC-GATA164A09, DXS7132, DXS7133, DXS9895, DXS9898, DXS6789, DXS6795, were screened. On each locus 5 – 11 alleles were noted, and DXS6789 proved the most polymorphic locus containing 11 alleles. No differences in the allelic distribution patterns of male and female subjects were found. Among female 4 – 35 different genotypes were noted with heterozygosity ranging 0.43 to 0.82.

In total, 29 cases of mutation were identified in 25 families covering all loci examined. The DXS6803 locus showed the highest mutation rate. In two families mutations were noted in three loci simultaneously. Most of the mutated cases were maternally derived and deletion type was slightly more prevalent than insertion type. The nine loci are on the same chromosome and it is safe to use haplotype rather than the usual multiplication rule for statistical analysis. Many cases of recombination were also noted, that is, parents transmitted one haplotype to their first child and another haplotype – haplotype with a different combination of alleles, but not a mutation – to the second child. Recombination occurred in every loci and no hot spot within different loci was apparent. It would appear that discrimination between the exclusional case and the inclusional case with mutation or recombination is very difficult with X-chromosomal haplotype only. Further detailed data will be presented.