

Criminalistics Section - 2008

B3 Evaluation of Eight X-Chromosomal STR Loci in Japanese Population

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The goal of this presentation is to discuss research involving eight X-chromosomal short tandem repeats. The X-chromosomal short tandem repeats (STRs) have recently been recognized to be useful tools in forensic medicine and anthropological studies for human identification as well as the distinctive properties of inheritance of the X- chromosome are responsible for its importance in population genetic studies. Features of X-chromosomal inheritance that are relevant to forensic casework will be discussed on the basis of empirical data, kinship, and paternity testing, mainly in deficiency paternity cases when the disputed child is a female. The goal of this study was to investigate the allelic frequency distribution of eight STRs on the X chromosome using the Mentype® Argus X-8 PCR amplification kit (Biotype AG, Germany), and evaluate the utility of this system in forensic medicine for the Japanese population.

This presentation will impact the forensic community by demonstrating the genetic evolution of eight X-chromosomal short tandem repeats (STR) DXS8378, HPRTB, DXS7423, DXS7132, DXS10134, DXS10074, DXS10101 and DXS10135 were fist reported in the Japanese population.

The X-chromosomal short tandem repeats (STRs) have recently been recognized to be useful tools in forensic medicine and anthropological studies for human identification as well as kinship and paternity testing, mainly in deficiency paternity cases when the disputed child is a female. The distinctive properties of inheritance of the X-chromosome are responsible for its importance in population genetic studies. Features of X-chromosomal inheritance that are relevant to forensic casework will be discussed on the basis of empirical data. In the cells of healthy human females, the X-chromosome is present as a homologous pair and resembles autosomes in this respect. The genetic evolution of eight X-chromosomal short tandem repeats (STR) DXS8378, HPRTB, DXS7423, DXS7132, DXS10134, DXS10074, DXS10101 and DXS10135 were examined in a sample of 353 unrelated males and females from the Japanese

population. Multiplex PCR amplification was performed using the Mentype $^{\circledR}$ Argus X-8 PCR amplification kit. The amplified PCR products were resolved and detected by capillary electrophoresis using the ABI PRISM 310 Genetic Analyzer. Allele frequencies of eight X-STR loci were calculated separately for males and females, and exact tests demonstrated no significant deviations from Hardy-Weinberg equilibrium. On the investigated kinship cases (30 family trios), no mutation was detected. Heterozygosity values ranged from 0.4419 (DXS7432) to 0.9269 (DXS10135), PIC ranged from 0.3805 (DXS7432) to 0.9222

(DXS10135). The combined power of discrimination (PD) for eight X-STR loci in males and females were 0.9992634 and 0.9999998, respectively. The eight X-STR loci form a new polymorphic marker system with great discrimination capacity for Japanese population.

X Chromosome, Short Tandem Repeat, Japanese Population