A STUDY OF RECOMBINATION BETWEEN 15 X CHROMOSOMAL SHORT TANDEM REPEAT MARKERS IN MULTIGENERATIONAL FAMILY PEDIGREES

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X chromosomal short tandem repeat (X STR) markers have recently been recognized as useful tools to supplement traditional kinship testing in the forensic setting. Development of assays allowing the multiplex detection and analysis of various combinations of X STRs has spawned numerous publications reporting the standardization of repeat structure and distribution of allele frequencies in a number of populations across the globe. However, far fewer studies have been published exploring the practical implications of utilizing markers located on a single chromosome.

According to the 1991 report of the International Society for Forensic Genetics (ISFG; formerly ISFH (Hameogenetics)) relating to the use of DNA polymorphisms in paternity testing, questions of independent assortment be addressed for any forensic marker system. For autosomal STRs, this ensures that the product rule can be used to multiply individual marker frequencies together to determine the overall rarity of a profile. It does not preclude the use of linked markers, however. Y chromosomal STRs, for example, are linked to one another and are considered together as a group called a haplotype. Haplotype frequencies are measured directly from population data, and the counting method is used to determine the rarity of the profile. It follows that X chromosomal STRs may require a combination of the two techniques: the organization of several physically close markers into linkage groups, forming haplotypes, whose frequencies could then be multiplied together once independent assortment of the groups or "blocks" was established. Early linkage studies produced a map of the X chromosome that divided 16 X chromosomal STRs into four linkage groups and most subsequent studies have employed this model when considering markers to include in novel multiplexes and the only currently available commercial X STR kit, QIAGEN's Investigator™ Argus X-12 kit.

At the Armed Forces DNA Identification Laboratory, two multiplexes consisting of a total of 15 X STR markers (DXS6789, DXS9902, DXS7132, DXS7130, DXS6795, DXS10147, DXS8378, DXS7423, HPRTB, DXS101, DXS7424, GATA31E08, GATA172D05, GATA165B12, and DXS6803) have been characterized and allele frequencies determined for several different populations. In order to evaluate the organization of these markers into the four proposed linkage groups, 58 families (832 individuals) satisfying the requirements of linkage study (multiple generations and offspring) have been acquired and investigated. Here, the authors will present the results of this recombination study.

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