

Population Genetics of DNA Typing and Applications to Forensic Case Work

(DNA Typing의 집단유전학과 법의 사건에의 적용)

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The DNA typing methods have substituted for the traditional blood typing in the forensic science. The surprising reliability enabled DNA evidence to be used for appointment of real criminal and powerful court evidence. Initially, the principles of DNA typing with restriction fragment length polymorphism were discovered by Jeffreys in 1985.

Nowadays, most of countries uses PCR based amp-FLP (amplified fragment length polymorphism) techniques and DNA sequencing even though United States or Canada still stick to RFLP in part in spite of so much disadvantages.

The majority of DNA typing includes amp-FLP in which VNTR (variable number of random repeat) or STR (short tandem repeat) loci of each chromosome be analyzed. As they show typical length variations, we can classify the genotypes by electrophoresis. Dozens of VNTR and hundreds of STR loci have been discovered so far, some of them can be selected as good forensic markers which are highly polymorphic in Korea through validation studies and random Korean population typing. Besides them, there are some useful forensic tools using PCR. For example, sequencing of mitochondrial amplification product can overcome the fatal limitations of amp-FLP. It enables the personal identification from even extremely degraded or aged (ancient) specimen on which conventional STR typing does not work. The accurate genotype of ABO antigen of each person is clarified by amplification followed by restriction enzyme analysis of ABO related glycosyl transferase gene. Using amplification of amelogenin gene region located at sex chromosome, the gender of unidentified blood stains or body remains can be disclosed. Case work includes mainly violent crimes (rape, homicide etc.), paternity tests and identification of missing persons generated from disaster like the breakdown of Sampoong Department Store.

In spite of the surprising reliability, to be accepted perfectly by the court, DNA evidence should be confirmed by some statistical values like random matching

probability (MP) or likelihood ratio (LR) by which court decides whether the appointed suspect by DNA evidence could be real criminal compared with circumstantial evidences. The accurate MP or LR value calculations are based on the construction of Korean random population data of corresponding VNTR or STR locus. As it is known that allelic distributions of VNTR and STR loci are quite different from race to race and even within a race, show slight but statistically significant deviations among reproductively isolated subpopulations or countries, each country should have its own population databases which satisfy Hardy-Weinberg equilibrium.

It is needless to say DNA typing brought turning point in forensic science. But, its comparative short history suggests us some future tasks. Each typing laboratory should have own standard protocol for the appropriate quality control. It is desirable to unify the marker loci among laboratories by which the information could be shared. The more rapid and reliable analysis method should be continuously developed.