B3 Individual Identification for Bones More Than 60 Years Old Using Autosomal SNPs on an Affymetrix® Chip

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After attending this presentation, attendees will understand the usability of Single Nucleotide Polymorphisms (SNPs) for individual identification in cases with aged bone samples as well as the basic principles involved in the use of resequencing arrays through practical analysis of case work using an Affymetrix® chip.

This presentation will impact the forensic science community in cases such as mass disasters by introducing a promising DNA typing tool. Short Tandem Repeat (STR) typing is typically performed to estimate genetic relationships, such as paternity tests; however, the limited number of STR markers, even though expanded more than previously before, is not sufficient enough in some cases. For example, it is hard to get full STR alleles in degraded samples such as bone or enough differentiation in very closed genetic relationships like those in an isolated population where the STR markers are only now being analyzed. In these cases, a supplementary tool is necessary to increase the power of matching probability with additional markers. SNP analysis is promising in this respect as it can be designed to be short for easy amplification, even in degraded samples, and can easily be multiplexed.

A resequencing array containing 169 autosomal SNP markers, which are distributed throughout the autosome except for chromosome 21, was used in this study on an Affymetrix® platform. The DNA chip analysis was applied to 14 identification cases involving bones that had been buried when the Jeju 4.3 incident occurred in South Korea from 1948 to 1954. Each case was comprised of recovered bone from a putative father and blood from an individual believed to be their child or sibling. For each analysis, the 169 SNP markers were applied to multiplex Polymerase Chain Reaction in a tube, then hybridized to probes on a chip. The statistical parameters, such as cumulative matching probability or cumulative identity index, were represented on an Identity by Descent (IBD) basis.

When comparing markers among the typed SNPs, the resulting data showed approximately 50 to 125 SNPs were shared in pairs. The genetic relationship was confirmed in all of the identification case pairs with over 99.99% of cumulative matching probability. In addition, no instances of discordance of STR alleles in a pair were found.

This study demonstrated a successful evaluation of genetic relationships was possible on challenging bone samples using autosomal SNP marker DNA chip resequencing results. Given the outcome of this study, it is expected that this test could be applicable to forensic samples as a complementary tool.

Resequencing, Affymetrix® Chip, Single Nucleotide Polymorphism