

PRODUCTION OF CUSTOMIZED RESEQUENCING ARRAY FOR SNP BASED HUMAN IDENTIFICATION

Hyung Jin Yu, Jisung Han, Yoon Soo Kim, Jong-Eun Lee, Dept. of Research & Developments, DNALink Inc.

Human identification technology first began in 1985 when DNA profiling and DNA fingerprinting were discovered. Since then, this technology has gradually improved to the current widespread method of analyzing STR (Short Tandem Repeat) markers. Despite the high accuracy and advanced analysis methods that are available today for human identification, confirming familial relationships is still impeded by the possibility of false positives due to the high rate of genetic mutations ($10E-3$) along with the hardship of obtaining quality result from degraded DNA samples.

In order to overcome such obstacles, new analysis techniques that utilize SNP (single nucleotide polymorphism) information have emerged. SNP is a DNA sequence variation occurring when a single nucleotide differs between individuals. By analyzing these genetic variations in the genome, SNP allows for many ways to accurately distinguish among individuals and groups.

Traditionally, a large quantity of DNA was needed in order to genotype proportionally large number of SNPs. However, DNA Link's AccuID (SNP based Personal Identification Technology) combines Affymetrix resequencing array technology and multiplexing PCR technology to genotype hundreds of SNP markers in a single experiment. It consists of markers for SNPs that are ideal for human identification as well as sex determination. Specifically, the system provides the genotyping of 169 human identification markers and 12 Y-chromosome sex markers in a single experiment run with small amounts of DNA as low as 50 pg. In addition, the system is set up to verify each marker 3 times, which significantly increases the accuracy of data. If the sample quality is low and not all 181 markers are able to be genotyped, using just 50 AccuID markers will match the error rate of the STR markers.

The series of AccuID to be released in the future will employ more markers, such as race and phenotypes (eye and hair color), to expand into broader applications and create whole new possibilities to the world of forensic science.