DEVELOPMENT AND VALIDATION OF A METHOD FOR PAIRWISE KINSHIP ANALYSIS USING HIGH-DENSITY SNP MICROARRAY

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Analysis of genome-wide single nucleotide polymorphisms (SNPs) data allows determination of the amount of hereditary information shared between two individuals, and could be useful for kinship analysis in forensic genetics, even for distant relationships. In this study, we developed a new method for pairwise kinship analysis using high-density SNP genotyping. We defined "index of chromosome sharing" (*ICS*) calculated using 174,254 SNP loci typed by a SNP microarray and by the genetic length of shared segments from the genotypes of two individuals. To investigate the expected *ICS* distributions from the first- to fifth-degree relatives and from unrelated pairs, we used computationally generated genotypes considering the effect of linkage disequilibrium (LD) and recombination. These distributions were used for calculating the likelihood ratio (LR) in pairwise kinship analysis, without allele frequencies and haplotype frequencies. Therefore, LD need not be considered for calculating LR or posterior probability. For validating this method, we collected buccal swabs from actual sample pairs including those of the first- to fifth-degrees of relations and genotyped all the samples using the HumanCore-24 BeadChip kit. Using this method, all actual sample pairs showed significantly high LR values (i.e., $\geq 10^8$). Therefore, we could distinguish distant relationships (up to the fifth-degree) from unrelated pairs based on LR. Additionally, our results indicated that this microarray showed high reproducibility (> 99.8%). Genotyping data of good quality (i.e., genotyping success rates > 99%) were obtained using as little as 10 ng DNA extracted from buccal swabs. Thus, our method greatly improves the pairwise kinship analysis of distant relationships and can be applied to actual forensic cases involving identification of disaster victims or missing persons.