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Usefulness of direct sequencing of pooled DNA for SNP identification and allele-frequency determination compatible with a common disease/common variant hypothesis

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ABSTRACT. We examined the efficiency of direct sequencing of pooled DNA for developing common single nucleotide polymorphisms (SNPs) and its accuracy for estimating allele frequencies. A pool of 200 control DNAs was established and was used for developing SNPs and estimating minor allele frequencies (MAF). The sensitivity of the pooled DNA method for successfully detecting an SNP with an MAF >0.01 listed in the database was approximately 0.7; it was particularly efficient for detecting SNPs with MAF >0.1, which is compatible with the common disease/common variant hypothesis. The mean difference between the estimated and the observed MAFs was 0.03 ± 0.023. The pooled DNA method identified four additional SNPs, for which the allele frequency

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information was not available in the database. The pooled DNA method is a cost- and time-effective tool for both qualifying and quantifying SNPs with considerable accuracy, and it can be particularly useful for dissecting the common disease/common variant hypothesis; this represents a best-case scenario for large-scale association mapping.

Key words: DNA; Pool; Single nucleotide polymorphism; Allele frequency

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