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Positional effects of polymorphisms in probe-target sequences on genoplot images of oligonucleotide microarrays

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ABSTRACT. Single nucleotide polymorphisms (SNPs) present in probe-target sequences (SPTS) have been shown to be associated with abnormal genoplot images. We explored the effects of SPTS positions on genoplot images using a data set from a genome-wide association study typed on an Illumina Human Hap300 platform. We screened the physical genomic positions of 308,330 autosomal probes to identify SPTS candidates deposited in dbSNP. The genoplot images across 293 individuals were inspected further in SNPs bearing an SPTS candidate. We identified 35,185 SNPs bearing a single SPTS candidate, including 264 SNPs showing abnormal genoplot images. The frequencies of SPTS at distances within 10 bases from the target SNP were significantly higher in the 264 SNPs showing abnormal genoplot images, than in the remaining 34,921 SNPs (49.62 *vs* 12.87%; Fisher exact test; $P = 2.2 \times 10^{-16}$). Of these 264 SNPs, we randomly selected 20 SNPs and resequenced them in 97 individuals. An SPTS within 10 bases of the target SNP was confirmed in all 20 SNPs, except for one SNP with a small deletion (7 bases) in the probe-

target sequence. Taken together, these results suggest an association of a proximal SPTS with an abnormal genoplot image, which could result in spurious genotype detections, highlighting the importance of minimizing systematic errors in microarray experiments.

Key words: Probe-target sequence; Genoplot image; Positional effects; Oligonucleotide microarray

