

Genotyping of human SNPs using single base extension-tag array methods

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Genotyping large numbers of human single-nucleotide polymorphisms (SNPs) in appropriate samples leads to insights into complex genetic traits including many common human diseases¹⁻³⁾. Here, we describe a method for parallel genotyping of SNPs, called single base extension-tag array on glass slides, SBE-TAGS. SNPs are genotyped by single base extension (SBE), using bifunctional primers carrying a unique sequence tag in addition to a locus-specific sequence⁴⁻⁵⁾. Marker-specific primers are used in PCR amplifications of genomic regions containing SNPs. The amplification products are used as templates in single base extension (SBE) reactions. The SBE primers, terminating one base before the polymorphic site, are extended in the presence of biotin labeled dideoxy NTPs, and hybridized to the tag array. After hybridization, the arrays were stained with streptavidin R-phycoerythrin solution to generate the fluorescence signal.

References

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