Phenotype Variations of Polymorphic Sites: Genotyping against Haplotyping

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Since nucleotide modifications including Single Nucleotide Polymorphisms (SNPs) may influence gene phenotypes thus, their studies highlight the distinctive features in pathogenesis of diseases. The reports on prediction of sequence profiles and patterns, as indicated in Prosite database (http://prosite.expasy.org), suggest that the function of elements and motifs is related to cumulative effect of the conserved signatures so that the polymorphic changes could affect inter- and intra molecular interactions (Kd value) leading to phenotype variations. Additionally, self-assembling proteins on several separate nucleotide elements shows that the phenotype variations may be depend on the function of other involved elements. Our knowledge of the involvement of numerous elements on the gene phenotype is limit. Furthermore, in most genotype reports obtained from multi SNPs studies, the heterozygote distribution of polymorphic sites on homologue chromosomes is not identified so that, they could not exactly show the phenotype variations. Theoretically, the number of their two-allele haplotypes can be estimated n! (n indicates the number of polymorphic sites), when the linkage failure exists between the sites. Thus, the phenotype reports based on the genotypes without consideration of haplotypes are not exactly estimated.

The haplotyping methods are able to determine the allele positions on the homologue chromosomes. Some of these methods estimate statistically the allele linkage between polymorphic positions (PHASE software) [1]. The precision of these methods are limit since the substitution rate, gene region and, tolerance level as considered from another so that, the phenotypes are exactly related to cumulative effects of the modified sites. These effects could be considered with the haplotyping techniques, usable in epidemiological studies. The progression in the direct procedures could markedly highlight the role of haplotypes in pathogenesis of diseases.

References


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