

Evolutionary-based Haplotype Association Studies

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Abstract

With the recent advancement in biotechnology and the completion of human genome sequence, many large-scale association studies have been conducted to identify the genetic region associated with the disease of interest. A common choice of the genetic marker is single nucleotide polymorphism, SNP. However, the number of SNP markers involved in this type of studies is usually large. When all markers are tested simultaneously, it runs into the problem of multiple testing. Therefore, using the fact that nearby SNPs are often correlated or appear together, it may be more efficient to consider haplotype as the unit in analysis. For instance, one can compare the haplotype frequencies between case and control groups. When the haplotype block is long and many haplotypes are possible in the region under study, one usually disregards those haplotypes with small frequencies. Others may prefer to group those minor haplotypes. Here we propose to reorganize these haplotypes based on coalescent theory. We will discuss what the theory is, and how it is applied to the association studies using case-control and family data. (This is a joint work with Mei-Hsien Lee and Jung-Ying Tzeng.)