Variable Set Enrichment Analysis in Genome-Wide Association Studies

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GEMS classroom, 3rd Floor in Shriner's Building
Coffee, water, and cookies will be provided

Abstract

Complex diseases such as hypertension are inherently multifactorial and involve many genetic and non-genetic factors of mild to minute effect sizes. A genome-wide association (GWAS) study typically tests hundreds of thousands of single nucleotide polymorphisms (SNPs), and offers the opportunity to evaluate the aggregated effects of many genetic variants with effects too small to be detected individually. I will present a new test for such aggregated effects of genes (or SNPs) linked by biological functions and known pathways. The new method (called variable set enrichment analysis, VSEA) extends the gene-set enrichment analysis (GSEA) of global gene expression by properly removing the effect of variable gene sizes when aggregating and by allowing for flexible analysis units (e.g., SNP sets). I will briefly describe the R package to perform real data analysis. The power of VSEA will be demonstrated by simulation modeling complex multi-loci interactions as well as real GWAS studies.