

CPH Seminar in Precision Medicine

FunSPU: an adaptive multiple functional annotations-based association test of whole genome sequencing data

Peng Wei, Ph.D.

Associate Professor, Department of Biostatistics
The University of Texas MD Anderson Cancer Center

Whole genome sequencing-based association testing of complex traits is challenging due to the large number of rare variants, many of which are non-trait-associated neutral ones. External biological knowledge, such as functional annotations based on the ENCODE and Roadmap Epigenome Project, may be helpful in distinguishing causal rare variants from neutral ones. However, each functional annotation can only provide certain aspect of the biological functions. Our knowledge to select the informative annotations a priori is limited while incorporating non-informative annotations will introduce noise and lose power. We propose the FunSPU test, a multiple functional annotations-based sum of powered score test that is adaptive at both the annotation and variant levels, thus maintaining high power even in the presence of no informative annotations. We conducted simulations to demonstrate the superior power of FunSPU compared with some existing methods that either ignore biological information or only consider a single functional annotation. We also illustrate our proposed test using the UK10K whole genome sequencing data based on six genome-wide functional annotations and identified several novel genomic loci that were missed by existing rare variant association tests. This is joint work with Yiding Ma.

Friday, April 28 2017. 12p – 1p. UCT.1414

Susan.Rojas@uth.tmc.edu

713.500.3654

 #SBMIseminar

