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Do haplotype tests gain power than collapsing tests in general pedigree-based association studies for detecting rare variants?

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Abstract

In recent years, the rare variants become more and more concerned with the advent of next-generation sequence (NGS) technology. Many rare variants are found to be causal in human disorders including psychiatric disorders, cardiac vascular diseases and cancer. The typical strategy is to collapse the rare variants as used in burden tests when analyzing the rare variants. In this manuscript, we focus on detecting the rare variants using pedigree data. Recently, a weighted haplotype test was considered in case-control data by Li et al. (2010) which showed greater power than the rare variants collapsing test in their simulation since haplotypes can carry some information of ungenotyped rare variants. Therefore, in this manuscript, we aimed to investigate the performance of haplotype collapsing tests in family data. As the PDT test, the proposed haplotype based PDT test (hPDT) considers the difference in the numbers of the transmitted and untransmitted haplotypes from parents to affected siblings and the difference in the numbers of haplotypes between affected and unaffected siblings. In simulation study, the forward evolutionary simulation tool *ForSim* is used to simulate genotypic data in pedigrees. To investigate the power, as a comparison, the rare variants based PDT test (rvPDT) is also considered in simulation.

Biography

Guo has completed her Ph.D. at the age of 30 years from Hong Kong University and postdoctoral studies from Ohio State University and Case Western Reserve University. She is a research fellow in Unit on Statistical Genetics in National Institute of Mental Health. She has published more than 10 papers in reputed journals.