



Application number/Title: 41722 - Statistical Method Development for Phenome-Wide Association Studies

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Keywords provided by the Applicant PI to describe the research project:

complex-diseases, gwas, multiple-phenotypes, phewas, statistical-method

Application Lay Summary:

In this project, we will develop new statistical methods to find genes responsible for complex human diseases and apply these methods to Biobank data sets. Over the last decade, genome-wide association studies (GWAS) have been widely performed to identify genetic associations for many complex diseases. A common limitation of GWAS is that they focus on only a single disease. Emerging evidence has shown that pleiotropy, the phenomenon of one genetic variant affects multiple diseases, is widespread in complex human diseases. Therefore, joint analysis of multiple diseases within a cohort presents an attractive alternative to single disease analyses and may provide new insights into the etiology of human diseases. Therefore, in this project, we plan to develop novel statistical methods to test the association between a genetic variant with a large number of diseases jointly. We plan to work on this project in the coming three years. We will publish the statistical methods, computational tools and biological findings from the data analysis and make the research results publicly available. Discovering novel genetic variants contributing to complex diseases may aid in improving diagnosis, treatment, and prevention, and have significant public health benefits.