The University of Chicago
Department of Statistics

MASTER'S THESIS PRESENTATION

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Methods for Detecting Trait Association with X-chromosome Markers in Pedigree Data

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ABSTRACT

We consider the problem of detecting association between a binary trait, such as a disease, and a single-nucleotide polymorphism (SNP) on the X-chromosome, when individuals are related with known relationships in a case-control study. Many statistical methods have been developed for case-control association testing. The method proposed here is based on the M_{QLS} test (Thornton and McPeek, 2007) for association between a binary trait and an autosomal SNP, for samples containing related individuals. The M_{QLS} test has been demonstrated to improve power substantially over previous tests, while retaining a computational simplicity that makes it useful in association studies in arbitrary pedigrees. Hence, we focus on extending the M_{QLS} test for autosomes to the X-chromosome. Under the null hypothesis of no association between the trait and the SNP, the standard X-chromosome inheritance model implies that males and females are expected to have the same allele frequency. Under the null hypothesis, covariance between the genotype variables at an X-chromosome locus for any pair of individuals can be calculated recursively using the pedigree information and assuming that Hardy-Weinberg equilibrium holds in the founders. This model setup ensures that the M_{QLS} test for autosomes can be extended to the X-chromosome. Finally, we focus on application of the new method to X-chromosome data from a genome-wide association study with related individuals.

Information about building access for persons with disabilities may be obtained in advance by calling Sandra Romero at 773.702-0541 or by email (sandra@galton.uchicago.edu).