



THE UNIVERSITY OF CHICAGO

Department of Statistics

DISSERTATION PROPOSAL

JOELLE MBATCHOU

Department of Statistics
The University of Chicago

Permutation-Based Methods for Assessing Significance in Genetic
Association Studies with Binary Traits and Related Individuals

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ABSTRACT

One of the main goals of human genetics is to identify genetic risk factors for common, complex diseases such as type 2 diabetes. Some recently proposed association tests involve aggregating across variants in a gene or region and lead to test statistics with unknown null distribution, an issue which can be solved by using permutation to assess significance. In the presence of related individuals in the sample, we encounter lack of exchangeability among the phenotype as well as the genotype which prevents us from using a simple permutation approach. We motivate and propose several permutation-based methods for binary traits that generate either phenotype or genotype replicates and accommodate for samples containing related individuals.

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