



THE UNIVERSITY OF
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Department of Statistics
DISSERTATION PROPOSAL

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Rare Variant Association Tests for Related Individuals

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ABSTRACT

Genetic association studies aim at identifying genetic polymorphisms that are associated with a disease or trait. Recent advances in next generation sequencing technologies necessitate the development of powerful statistical tests for rare variant associations. Existing approaches focus on combining variants across a genetic region and assume independence among sampled individuals, hence do not address relatedness in the sample.

We consider the problem of rare variant association tests for a sample of related individuals with a known pedigree. I will present our method closely connected to SKAT-O introduced by Lee et al (2012) for analysis of quantitative traits. Specifically, effects of rare variants within a genetic region or a functional group are modeled as random effects having a covariance structure with an unknown correlation coefficient. Optimal tests are then chosen across the nuisance correlation coefficients to maximize power. I will discuss simulation results to illustrate the performance of our method, as well as future directions of research to improve and generalize this approach.

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