



The University of Chicago
Department of Statistics

FIRST YEAR PHD PRESENTATION

SCOTT POWERS

Department of Statistics
The University of Chicago

**Assessing the Impact of Non-differential Genotyping
Errors on Rare Variant Tests of Association**

TUESDAY, May 22, 2012, at 4:30 PM

110 Eckhart Hall, 5734 S. University Avenue

ABSTRACT

We aim to quantify the effect of non-differential genotyping errors on the power of rare variant tests and identify those situations when genotyping errors are most harmful. We simulated genotype and phenotype data for a range of sample sizes, minor allele frequencies, disease relative risks and numbers of rare variants. Genotype errors were then simulated using five different error models covering a wide range of error rates. Even at very low error rates, misclassifying a common homozygote as a heterozygote translates into a substantial loss of power, a result that is exacerbated even further as the minor allele frequency decreases. While the power loss from heterozygote to common homozygote errors tends to be smaller for a given error rate, in practice heterozygote to homozygote errors are more frequent and, thus, will have measurable impact on power. Error rates from genotype-calling technology for next-generation sequencing data suggest that substantial power loss may be seen when applying current rare variant tests of association to called genotypes.

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