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Practice Job Talk Presentation

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Testing Untyped SNPs in Case-Control Association Studies with Related Individuals

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ABSTRACT

Genome-wide association studies are widely used to try to identify genetic factors influencing human complex disease. In current high-throughput genotyping platforms, only a subset of all genomic variants are genotyped. However many of the untyped variants may be well-predicted from the typed variants, with information on the joint distribution of typed and untyped variants available from an external reference panel such as HapMap. Incorporation of such external information can allow one to perform tests of association between untyped variants and disease, thereby making more efficient use of the available genotype data. A missing-data problem results from the fact that only genotype and not haplotype data are available. A further complication is that casecontrol samples may contain related individuals, resulting in dependence among sampled genotypes. I develop a method (ATRIUM) to test for association between untyped SNPs and disease, based on genotype data on associated markers with missing data allowed, where the test is applicable in samples that contain related individuals, assuming that the individuals are outbred and that pedigree information is available. ATRIUM is a quasilikelihood score test that allows for both missing and dependent data. Because the external reference panel information is used only to select the direction in which to perform the 1-df test of association with an untyped SNP, our test maintains the nominal type I error rate even when the external LD information is misspecified. As a result, our test is robust to mismatch between the case-control and reference samples. I apply the method to test for association with type 2 diabetes in a sample containing related individuals.

This is a joint work with Dr. Mary Sara McPeek.

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