Assessing the Effects of Two Epigenetic Factors on Complex Genetics Traits

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Abstract

Genomic imprinting and maternal effects are two epigenetic factors that are being explored to assess their effects on the etiology of complex diseases. Although methods have been proposed for case-parents triads and case-mother/control-mother pairs designs, they are not amenable to extended families. Further, existing methods are full-likelihood based and have to make strong assumptions concerning mating type probabilities (nuisance parameters). In this talk, I will focus on Likelihood approaches for detecting Imprinting and Maternal Effects (LIME) simultaneously using family data. In particular, I will discuss LIME-ped, which uses extended pedigrees and introduces a novel concept called "conditional mating type" to avoid making the Hardy-Weinberg equilibrium assumption. I will also discuss LIME-mix, which matches the case families with control families of the same structure and stratifies them according to the familial genotypes. This enable us to derive a partial likelihood that is free of nuisance parameters. I will show simulation results to illustrate power gain with LIME-ped by using extended pedigrees and demonstrate robustness of LIME-mix under a variety of settings.

Keywords: Genomic imprinting; Maternal effect; Partial likelihood; General pedigrees; Case-parents/control-parents triads; Case-mother/control mother pairs.

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