

Robust Partial Likelihood for Detecting Imprinting and Maternal Effects

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ABSTRACT. Genomewide association studies can only explain a small proportion of phenotypic variation. Epigenetic effects have been pointed out to be one plausible source of the missing heritability. Numerous statistical methods have been developed to explore two important epigenetic factors: genomic imprinting and maternal effects. Most of the methods, however, can only model one of these two confounded epigenetic effects or make strong yet unrealistic assumptions about the population to avoid overparameterization. In this talk, I will present a partial Likelihood method for detecting Imprinting and Maternal Effects jointly without making assumptions about the nuisance parameters. This method can be applied to both case-control family data and discordant sib-pair design. By matching affected and unaffected probands and stratifying according to their familial genotypes, a partial likelihood component free of nuisance parameters can be extracted from the full likelihood and alleviates the need to make assumptions about the nuisance parameters. Theoretical analysis shows that the maximum partial likelihood estimators based on the partial likelihood method are consistent and asymptotically normally distributed with closeform formula for computing information. Based on the information contents per individual of different study designs, our work offers a practical strategy and realtime information calculation for investigators to select the optimum study before data collection.