

Detection of cell-type-specific risk-CpG sites in epigenome-wide association studies

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Abstract: Classic approaches to association studies that link genomic features with phenotypes always either regress the phenotypes on the genomic features or regress the genomic features on the phenotypes. However, in epigenome-wide association studies, the measured signals for each sample are a mixture of methylation profiles from different cell types. Previous methods for association detection claim whether a cytosine-phosphate-guanine (CpG) site is associated with the phenotype or not at aggregate level with the classical regression methods and can suffer from low statistical power. Here, we propose a statistical method, High REsolution (HIRE), which not only improves the power of association detection at aggregate level as compared to the existing methods but also enables the detection of risk-CpG sites for individual cell types.