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TITLE: Using imputed genotype data in the joint score tests for genetic association and gene-environment interactions in case-control studies
ABSTRACT BODY:

Abstract Body: Genome-wide association studies (GWAS) are now routinely imputed for untyped single nucleotide polymorphisms (SNPs) based on various powerful statistical algorithms for imputation trained on reference datasets. The use of predicted allele counts for imputed SNPs as the dosage variable is known to produce valid score test for genetic association. In this paper, we investigate how to best handle imputed SNPs in various modern complex tests for genetic associations incorporating gene-environment interactions. We focus on case-control association studies where inference for an underlying logistic regression model can be performed using alternative methods that rely on varying degree on an assumption of gene-environment independence in the underlying population. As increasingly large-scale GWAS are being performed through consortia effort where it is preferable to share only summary-level information across studies, we also describe simple mechanisms for implementing score tests based on standard meta-analysis of "one-step" maximum-likelihood estimates across studies. Applications of the methods in simulation studies and a dataset from GWAS of lung cancer illustrate ability of the proposed methods to maintain type-I error rates for the underlying testing procedures. For analysis of imputed SNPs, similar to typed SNPs, the retrospective methods can lead to considerable efficiency gain for modeling of gene-environment interactions under the assumption of gene-environment independence. Methods are made available for public use through CGEN R software package.

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