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Speaker:
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Title:
A novel region-based Bayesian approach for genetic association with next generation sequencing (NGS) data coverage.

Abstract:
The discovery of rare genetic variants through Next Generation Sequencing (NGS) is becoming a very challenging issue in the human genetic field. We propose here a novel region-based statistical test based on a Bayes Factor (BF) approach to assess evidence of association between a set of rare variants located on this region and a disease outcome. Marginal likelihood is computed under the null and alternative hypotheses assuming a binomial distribution for the rare variants count in the region. A Beta distribution or a mixture of Dirac and Beta distribution is specified for the prior distribution. The hyper-parameters are determined to ensure the null distribution of BF does not vary across genes with different sizes. A permutation test or False Discovery Rate (FDR) statistic are used for inference. Our simulations studies showed that the new BF statistic outperforms standard methods under most situations considered. Our real data application to a lung cancer study found enrichment for rare variants in novel genes.