Assessing the impact of missing genotype data in rare variant association analysis

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Human genome resequencing technologies are becoming ever more affordable and provide a valuable source of data about rare genetic variants in the human genome. Such rare variation may play an important role in explaining the missing heritability of complex human traits. We implement an existing method for analyzing rare variants by testing for association with the mutational load across genes. In this study, we make use of simulated data from the Genetic Analysis Workshop 17 to assess the power of this approach to detect association with simulated quantitative and dichotomous phenotypes and to evaluate the impact of missing genotypes on the power of the analysis. According to our results, the mutational load based rare variant analysis method is relatively robust to call-rate and is adequately powered for genome-wide association analysis.

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