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TITLE: Do common and rare variants work together on the same gene to affect phenotype?

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ABSTRACT:
More and more evidence indicate that the rare variants could explain a part of the genetic variance. However, how the common variants and rare variants work together to control the phenotype is still unclear. How many associated rare variants collocated on the same gene with associated common variants? Or the rare variants rarely overlapped with the common variants as they have their own mechanism to affect phenotype?

To answer these questions, several comparisons among different GWAS methods were implemented in two datasets. One dataset is the exome-sequenced UK biobank data, which was used for the GWAS of rare variants and includes 49,960 samples and 10,448,724 SNPs. Another dataset is the imputed UK biobank DNA array data, which was used for the GWAS of common variants and includes 486,565 samples and 89,716,503 SNPs. MAGMA, SKAT, and burden test were used in GWAS analysis. There were 6 phenotypes including height, BMI, diabetes, heel bone density, blood pressure, and basal metabolic rate were used in the comparison.

The results indicated that there is no significant correlation among 90 comparisons between different methods in 6 phenotypes. Although it is too early to state that the rare variants and common variants are not closely related in gene-level, this result could be

the benchmark in the following research of the relationship of rare and common variants.

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