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Project Title

Comprehensive catalog of annotated genetic variations and reference haplotypes for genotype imputation to support COVID-19 research in Quebec

Project Summary

Genotyping array technology remains the most affordable option for large-scale population-based genome-wide association studies (GWAS). However, out of hundreds of millions of known genetic variations, a typical genotyping array platform assesses only ~1 million. The remaining millions of unassessed variations are inferred using the specialized statistical methods, known as imputation methods, from a collection of reference genomes. The quality of the imputed variations and subsequent analyses depends on the composition of the reference genomes. The statistical inference will be better when using reference genomes which best represent the local population. This study constructs the population-specific reference genomes from the Biobanque québécoise de la COVID-19 (BQC19). We will apply these reference genomes to CLSA genotype array data to improve the quality of imputed genetic variations. We will test the imputed variations for associations with COVID-19 status.

Keywords

genotype imputation, reference haplotypes, gwas, genetic association, HLA, COVID-19