Some Perspectives of Graphical Methods for Genetic Data

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Abstract

Recent initiatives have made genetic data on single-nucleotide polymorphisms (SNPs) in humans widely available. The association study between these SNPs and a host of measures in humans and other species has led to a vigorous development of analytical tools as with a great understanding of the genetic basis of common diseases. Among many aspects of the data analysis, there is a need to synthesise the graphical methods involved. I give a brief account of the background, provide examples in recent analyses, and draw attention to further work.

Specifically, the examples provided are from a number of aspects: 1. Phenotypic data. While this includes the usual summary statistics it may also be specific to genetic context such as pedigree-drawing. 2. Genotypic data. This includes plotting missing data, Hardy-Weinberg equilibrium (HWE), and the correlation between neighbouring SNPs (LD). 3. Assessment of population substructure and genotype-phenotype association. This includes scree plot, Manhattan plot, Q-Q plot, SNP-based summary plot and regional association plot. 4. Representation of pathways. Other examples may arise from study of power, meta-analysis and interactions. In addition, a comparison will be made between graphics from CRAN packages with popular standalone programs such as LD plot.