Abstract

Computational and Statistical Challenges of Genome-Wide Detection of Biomarkers Associated with Diseases and Agricultural Traits

Prof. Adam Kowalczyk

Centre for Neural Engineering, The University of Melbourne, Australia

Personalised medicine, direct to public genotyping services and precision agriculture depend critically on the ability to identify DNA regions in which there are causal mutations and/or variants that predict trait variation. With the impressive progress in genotyping technology in the last decade, the bottleneck has shifted from wet labs to bioinformatics: the statistical and computational analysis of data. State-of-the art bioinformatics is critical for resolution of the missing heritability challenge that has arisen from the fact that conventional analyses of genome-wide association studies have typically identified genomic markers that explain only a small fraction of the familial correlation/association of the trait. In particular, current approaches have shortcomings as attested to by the fact that they have generally failed to detect causal mutations, let alone the genetic interactions (epistasis) which are known must exist but are poorly understood.

The spectacular progress in practical computational technology finally makes feasible to address these challenges. This presentation will illustrate in a popular form the “cosmic” size of the challenge and discuss some novel computational and statistical methodologies designed to address this bottleneck.