Abstract/Session Information for Program Number 1486F

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Session Information

Session Title: Statistical Genetics and Genetic Epidemiology **Session Type:** Poster

Session Location: Exhibit Hall, Lower Level South, Moscone Center **Session Time:** Fri 7:00AM-4:30PM

Abstract Information

Program Number: 1486F **Presentation Time:** Fri, Nov 9, 2012, 3:15PM-4:15PM

Keywords: Statistical Genetics and Genetic Epidemiology, KW140 - population genetics, KW075 - genetic mapping, KW080 - genome-wide association, KW031 - computational tools, KW008 - bioinformatics

Abstract Content

Detection of identity by descent based on rare variants. *G. Povysil, G. Klambauer, S. Hochreiter* Institute of Bioinformatics, Johannes Kepler University Linz, Linz, Austria.

Identity by descent (IBD) between two individuals means that their alleles are identical because they were inherited from a shared common ancestor. Detection of IBD tracts is important for population genetics and association studies. IBD detection methods perform well for family studies where pedigrees are available and for common single nucleotide variants (SNVs). However, recent genotyping projects utilizing next generation sequencing comprise unrelated individuals and detect mostly rare variants. Currently, rare variants are of high interest in genetics because they are assumed to cause complex human diseases. However, their association with a disease is hard to detect as standard tests on rare variants yield low power. IBD mapping can be used to increase the power by two approaches. First, SNVs can be grouped based on IBD and subsequently their joint effect tested for disease association. Secondly, local genetic similarities between individuals can be measured by IBD and used for association tests like implemented in the sequence kernel association test (SKAT).

We compare two of the most commonly used IBD detection techniques, BEAGLE's fastIBD and PLINK, on simulated rare SNVs with implanted IBD tracts of different sizes and different proportions of minor alleles. Both methods miss a large proportion of short tracts and tracts that are tagged by few minor alleles. Overall fastIBD has higher power than PLINK, which is traded off against a higher false discovery rate. Besides correctly identifying IBD tracts, an exact estimation of IBD length and location is essential for identifying disease loci by IBD mapping. In this respect fastIBD systematically overestimates the length of IBD tracts while PLINK is very accurate. Our findings may help to choose an appropriate IBD detection method for studies involving rare variants. Summarizing, fastIBD finds almost all IBD tracts on the cost of many false positives whereas PLINK finds a smaller number of IBD tracts of which only a few are false positives.

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