PERSONAL GENOMICS

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1. Introduction

Our genetic identity not only determines our physical differences, but it also defines our susceptibility against diseases. Several groups are working on various methods to exploit the power of cost efficient sequencing technologies as well as more traditional genome analysis approaches (SNP microarrays, arrayCGH, etc.) to better perform genotype-phenotype associations, in particular to identify susceptibility to disease, and eventually diagnose disease at its early stages. The ultimate goal is to vastly improve the field of pharmacogenomics, which can broadly be defined as the study of the relationship between genotype and drug response and how the drugs affect our metabolism. The abundance of new sequence data gives many opportunities to advancing our understanding of how to optimize drug combinations for each individual's genetic makeup. The underlying computational tools for such studies analyze available sequence data to identify differences between a reference genome and high-throughput sequenced genomes and perform sequence oriented clustering and classification to obtain both normal and disease-related phenotype associations.

This session focuses on the development of novel computational methods in all aspects of Personal Genomics including genetic and epigenetic variation discovery, genotype-phenotype associations, indexing and cataloguing both normal and disease-related variation, exome capture and resequencing, and personalized medicine. This session has a broad target audience that includes algorithm developers working on sequence analysis, genomics researchers, pharmacogeneticists, and medical geneticists.

2. Session Summary

This session includes an invited talk, six reviewed oral presentations, and a tutorial. The studies presented in this session focus on the development of computational methods to analyze genomic data generated with various types of methods.

2.1. *Oral Presentations*

The following six talks will be presented at the Personal Genomics session:

- "Haplotype Inference from Single Short Sequence Reads Using a Population Genealogical History Model" by Jim Zhang and Yufeng Wu,
- "Multivariate Analysis of Regulatory SNPs: Empowering Personal Genomics by Considering Cis-Epistasis and Heterogeneity" by Stephen D. Turner and William S. Bush,
- "Visual Integration of Results from a large DNA Biobank (BIOVU) using Synthesis-View" by Sarah Pendergrass, Scott M. Dudek, Dan M. Roden, Dana C. Crawford, and Marylyun D. Ritchie,
- "Use of Biological Knowledge to Inform the Analysis of Gene-Gene Interactions Involved in Modulating Virologic Failure with Efavirenz-Containing Treatment Regimens in Art-Naïve ACTG Clinical Trials Participants" by Benjamin J. Grady, Eric C. Torstenson, Paul J. Mclaren, Paul W. De Bakker, David W. Haas, Gregory K. Robbins, Roy M. Gulick, Richard Haubrich, Heather Ribaudo and Marylyn D. Ritchie,
- "The Reference Human Genome Demonstrates High Risk of Type 1 Diabetes and Other Disorder" by Rong Chen and Atul J. Butte
- "Matching Cancer Genomes to Established Cell Lines for Personalized Oncology" by Joel T. Dudley, Rong Chen, and Atul J. Butte

We are excited by the breadth of research in the field of Personal Genomics, and are hopeful that our session will help bring together researchers in these areas. The six papers presented at our session were selected with the help of several reviewers, whose help we gratefully acknowledge.

3. Acknowledgments

We would like to thank all the authors who submitted their work to the Personal Genomics Session. We are also indebted to the anonymous reviewers who contributed their time and expertise to evaluate the submitted papers.