CIHR **STAGE**

Strategic Training for Advanced Genetic Epidemiology

International Speaker Series

Prof. Bruce Rannala

Professor, Genome Center and Department of Evolution and Ecology
University of California Davis

Statistical analysis of pooled samples for whole-genome case-control associations: A SNP GWAS of lung cancer susceptibility genes in the Northern Thai population

Friday
November 1, 2013
12:00 – 1:00 pm

The Hospital for Sick Children
CDIU Multimedia Theatre
Room 4132, 4th Floor, Elm Elevators
555 University Avenue, Toronto, ON

Abstract: Genome-wide association studies (GWASs) have been extensively applied in case-control studies aimed at identifying single nucleotide polymorphisms (SNPs) in the human genome that are linked to complex diseases including cancer. However, the newest chip-based assays that interrogate hundreds of thousands of SNPs and examine individual genotypes remain expensive and most GWA studies are therefore being conducted in developed countries. GWA studies of populations in the developing world may help to identify new variants associated with disease because many developing countries have unique population genetic compositions and environmental exposures that differ from developed countries. A promising strategy that allows geneticists to carry out cost-effective GWA studies in developing countries is the pooled GWAS in which DNA samples from cases and controls are separately pooled and genotyped on single chips. Results are presented from a pooled GWAS aimed at identifying SNPs influencing lung cancer susceptibility in the population of Northern Thailand (which has a high incidence of lung cancer relative to other regions). Our study used the Illumina Infinium Human660W Quad BeadChip. I will describe some of the statistics used for comparing SNP allele frequencies in cases versus controls as well as methods for eliminating chip- or pooling-based artifacts. About a dozen "SNPs of interest" were identified that appeared to differ in frequency between cases and controls with p-values ranging from 10^(-3) to 10^(-8). Several of the identified SNPs have been previously associated with cancers or are linked to genes with a known role in cancer. The total cost of this study was orders of magnitude less than an individual genotype-based study.

Profile: Bruce Rannala is a Professor in the Genome Center and Department of Evolution and Ecology at UC Davis. He previously held positions on the faculty of SUNY Stony Brook and the University of Alberta and is currently a guest Professor at the Beijing Institute of Genomics, China. He was the Canadian Institutes of Health Research Peter Lougheed Scholar in 2001 and was appointed as a Miller Professor at UC Berkeley in 2009. Bruce Rannala's principal research interests include statistical aspects of population genetics, human genetics and phylogenetic inference. In particular, he has contributed to the development of Bayesian methods for phylogenetic inference, methods for population genetic inference of fine-scale recombination rates and ages of mutations, and individual-based assignment methods for migration rate inference. Bruce Rannala received a B.Sc. in Zoology from the University of British Columbia in 1989 and a Ph.D. in Biology from Yale University in 1995. He was an NSERC postdoctoral fellow at UC Berkeley.

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