CIHR **STAGE**

Strategic Training for Advanced Genetic Epidemiology

Dr. Kimberly Siegmund

Associate Professor of Biostatistics
Department of Preventive Medicine, USC
Keck School of Medicine
University of Southern California

DNA Methylation Studies using Illumina HumanMethylation450 BeadArrays

Friday
March 1, 2013
12:00 –1:00 p.m.

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

Profile: Kimberly D. Siegmund is Associate Professor of Biostatistics in the Department of Preventive Medicine and Member of the USC Norris Comprehensive Cancer Center at the University of Southern California Keck School of Medicine. Dr. Siegmund received her Ph.D. in Biostatistics in 1995 from the University of Washington. Her primary research interest is statistical methods in epigenetics, where she has focused on developing methods for microarray signal processing, class discovery, and classification of DNA methylation data. She collaborates on studies of DNA methylation in cancers of the lung, colon, bladder and Alzheimer's disease, has co-authored a book chapter on epigenetics for the Handbook of Statistical Genetics (John Wiley & Sons, Ltd. 2007), and published a review article, "Statistical approaches for the analysis of DNA methylation microarray data" (Human Genetics, 2011). Presently, she and her colleagues are involved in the statistical analysis of Illumina BeadArray and high-throughput bisulfite sequence data generated at the USC Epigenome Center.

Abstract: Variation in the epigenome, the positioning of DNA-related modifications and structural features that inform the packaging of DNA, can confer a host of specialized functions to different cells with the same genome. DNA methylation is the most commonly studied epigenetic mark; its importance well-established in human development and disease. Presently, DNA methylation microarrays provide the most cost-effective means of high-throughput analysis. As with other types of microarrays that measure gene expression, genotype, or copy number variation, technical artifacts are a concern. I will introduce the Illumina HumanMethylation450 platform, discuss approaches to assess data quality, and present recently developed methods for signal processing. Specifically, I will discuss how to leverage 'hidden' information on the array to correct for background fluorescence, and describe a method to correct for bias from using two fluorescent dyes. The methods will be illustrated using replicate control and biological samples from HumanMethylation27 and HumanMethylation450 BeadArrays. I will conclude with a discussion of the variety of statistical approaches applied in DNA methylation association studies.

Housed at the University of Toronto Dalla Lana School of Public Health, CIHR **STAGE** is a training program in genetic epidemiology and statistical genetics funded by the Canadian Institutes of Health Research through the Strategic Training Initiative in Health Research program. Seminars are sponsored by The Hospital for Sick Children, the Samuel Lunenfeld Research Institute of Mount Sinai Hospital, the Ontario Institute for Cancer Research, the Department of Statistics of the University of Toronto, the Ontario Cancer Institute of the University Health Network, and the CIHR Institute of Genetics.

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