THE OHIO STATE UNIVERSITY
COLLEGE OF MEDICINE

Department of Biomedical Informatics

Integrative statistical approaches for the analysis of whole-genome sequencing data

Abstract: Continuous advances in massively parallel sequencing technologies make large whole-genome sequencing studies increasingly feasible, including the NHLBI Trans-Omics for Precision Medicine (TopMed) and UK Biobank studies. The analysis of such data is challenging due to the large number of rare variants in noncoding regions of the genome, our poor understanding of their functional effects, and the lack of natural units for testing (e.g. the analogue of genes in coding regions). In this talk I will describe some of our effort to address these challenges using statistical and computational approaches. In particular, I will discuss a scan statistic method to simultaneously detect the existence, and estimate the location of the association at genome-wide scale. Our method can incorporate a large number of functional annotations of genetic variants for improved power to identify the signals in noncoding regions. I will show applications to several datasets, including whole-genome sequencing data in the Simons Simplex Collection (SSC) and TopMed studies.

About the Speaker: Iuliana Ionita-Laza joined Columbia University in 2009, where she is currently an associate professor of biostatistics. Ionita-Laza's research area is at the interface between statistics, machine learning and genomics, with emphasis on the development of statistical and computational methods for problems arising in human genomics. Particular interests include the development of methods for whole genome sequencing data, and integrative statistical methods for different types of omics data. In addition to her methodological component, she is also involved in the application of these methods to sequencing studies for several complex phenotypes. Ionita-Laza received her PhD from the Courant Institute of Mathematical Sciences at NYU in 2006, and subsequently was a postdoctoral fellow in the Department of Biostatistics at Harvard University. She is currently an associate editor for Biometrics, and Statistics in Biosciences, and serves on the Genomics, Computational Biology and Technology study section.

Iuliana Ionita-Laza, PhD

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165 DHLRI