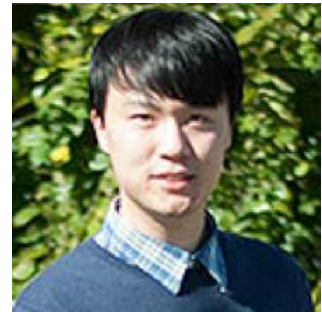


Statistics SEMINAR

Title: Integrative analysis of epigenetic and transcriptomic annotation data provides insights into the genetic basis of complex human diseases

Speaker: Qiongshi Lu

**Department of Biostatistics and Medical Informatics
University of Wisconsin-Madison**



Time & Place:

Wednesday, December 13, 2017

4pm, Room 133 SMI

Cookies & Coffee @ 3:30, Rm 1210

MSC

Abstract: Continuing efforts from large international consortia have made genome-wide epigenomic and transcriptomic annotation data publicly available for a variety of tissue types. Computational methods have also been developed to effectively synthesize these data into summary metrics and characterize the functional non-coding genome. In this talk, I will present a few examples to demonstrate how to integrate tissue-specific functional annotations in genetic association studies for complex diseases. Specifically, we use data from 127 different cell types to present an atlas of enrichment across 45 different complex traits. Additionally, we introduce a principled framework to perform powerful, annotation-stratified genetic covariance estimation. Finally, we present a new cross-tissue analytical framework to perform gene expression imputation as well as gene-level association test. An in-depth case study of several neurodegenerative diseases, including Alzheimer's disease (AD), Parkinson's disease (PD), and amyotrophic lateral sclerosis (ALS), will be discussed. Our analyses suggest a strong connection between neurodegeneration and genetic variants in monocyte-specific functional genome. We also show that the localization of SNPs to monocyte-functional regions is a pattern of inheritance shared between AD and PD. Finally, we discuss the shared and distinctive genetic architecture between AD and ALS. Overall, we show that integrated genome annotations at the single tissue level may be a valuable tool for understanding the etiology of complex human diseases.

