Indiana University-Purdue University Indianapolis

Department of Mathematical Sciences

Statistics Seminar

12:15pm—1:15pm, Tuesday, April 25, 2023
Zoom Meeting: Meeting ID: 845 0989 4694

Speaker: **Ming Li**
Department of Epidemiology and Biostatistics,
Indiana University Bloomington School of Public Health

Title: **Methylation Quantitative Trait Loci and the Risk of Congenital Heart Diseases**

Abstract:

DNA methylation may be regulated by genetic variants within a genomic region, referred to as methylation quantitative trait loci (mQTLs). The changes of methylation levels can further lead to alterations of gene expression and influence the risk of various complex human diseases, such as congenital heart diseases (CHDs). Detecting mQTLs may provide insights into the underlying mechanism of how genotypic variations may influence the disease risk. We investigated the cis-acting effects of genetic single nucleotide polymorphisms (SNPs) on local DNA methylation patterns within cardiac tissue samples and prioritized their contributions to CHD risk by leveraging results of CHD genome-wide association studies and their effects on cardiac gene expression. Through these studies, we also proposed a series of methylation random field methods to improve the detection of mQTLs. The proposal methods use beta distributions to characterize the bimodal and interval properties of methylation traits, consider multiple common and rare genetic variants within a genomic region to identify mQTLs, and allows flexible correlation structures between neighboring CpG sites. Our findings support the hypothesis that genetic variants may influence the risk of CHDs through regulating the changes of DNA methylation and gene expression.

Bio:
Dr. Ming Li is an associate professor in the Department of Epidemiology and Biostatistics at Indiana University Bloomington School of Public Health. He attained his PhD degree from Michigan State University in 2012 and was an assistant professor at University of Arkansas for Medical Sciences between 2012 and 2015 before joining IU in 2015. Dr. Li's research interests are in the field of statistical genetics and

genetic epidemiology, with a focus on the development of biostatistical methods and their application to complex human diseases, such as birth defects.