

Sept. 17: Jianxin Shi, Ph.D.: Characterizing the Genetic Basis of Methyloome Diversity in Histologically Normal Human Lung Tissue



SYNOPSIS:

Epigenetic modification such as DNA methylation plays a key role in disease susceptibility and gene regulation. However, the genetic regulation of DNA methylation in human is not fully appreciated. Based on the Environment And Genetics of Lung Etiology (EAGLE) study in the NCI's Division of Cancer Epidemiology and Genetics (DCEG), we performed a methylation-quantitative trait loci (meQTL) analysis in 210 histologically normal human lung tissues. We identified 34,304 *cis*- and 585 *trans*-meQTLs, a genetic-epigenetic interaction of surprising magnitude. These findings are replicated in both breast and kidney tissues and show distinct patterns: *cis*-meQTLs mostly localize to CpG sites outside of genes, promoters and CpG islands (CGIs), while *trans*-meQTLs are over-represented in promoter CGIs. meQTL SNPs are enriched in CTCF-binding sites, DNaseI hypersensitivity regions and histone marks. Importantly, four of the five established lung cancer risk loci in European ancestry are *cis*-meQTLs and, in aggregate, *cis*-meQTLs are enriched for lung cancer risk in a genome-wide analysis of 11,587 subjects. Thus, inherited genetic variation may affect lung carcinogenesis by regulating the human methylome. We developed a novel statistical algorithm and identified an association hotspot with replication in The Cancer Genome Atlas (TCGA) samples.

[Session details...](#)

BIO:

Jianxin Shi received a Ph.D. in Statistics from Stanford University in 2006. He then pursued a postdoctoral research fellowship at the Stanford University School of Medicine focusing his studies on the genetic mapping of psychiatric disorders from 2006 to 2009. Dr. Shi is currently a tenure track investigator in the Biostatistics Branch of the Division of Cancer Epidemiology and Genetics at the NCI. His research focuses on cancer genetics and genomics.

SUMMARY:

Topic: Characterizing the Genetic Basis of Methyloome Diversity in Histologically Normal Human Lung Tissue

Speaker: Jianxin Shi, Ph.D.

Date: Wednesday, September 17, 2014

Time: 11 AM – 12 PM EDT

You are invited to listen to Dr. Shi's presentation in Room 2W908 in the NCI Shady Grove Building on Medical Center Drive or via WebEx.

Presentation: A screen cast of the presentation will be available for viewing after the event on the [NCI CBIIT Speaker Series YouTube Playlist](#).

About the NCI CBIIT Speaker Series:

The National Cancer Institute (NCI) Center for Biomedical Informatics and Information Technology (CBIIT) Speaker Series is a bi-weekly knowledge-sharing forum featuring both internal and external speakers on topics of interest to the biomedical informatics and research communities. For additional information, including past speaker series presentations, visit the [CBIIT Speaker Series page](#).

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