



INTERDISCIPLINARY DATA SCIENCES CONSORTIUM

SEMINAR SERIES

Functional Interpretation of Human DNA Variants Using Existing Knowledge and Machine Learning

DNA sequencing technologies continue to make progress in increased throughput and quality, and decreased cost. Our ability to convert machine-generated variant calls, including single nucleotide variant (SNV) and insertion-deletion variants (indels), into human-interpretable knowledge has lagged far behind the will discuss the challenges we are facing to obtain functional interpretations for the DNA variants observed in human genome sequencing studies, especially for genetic epidemiology and medical genetics, and some of the works that are done in my lab to help to answer those challenges.

FEBRUARY 7, 2020

2:00-3:00PM

LOCATION: CPR 203

To learn more, visit:

<https://idscbigdata.com/>

<https://health.usf.edu/publichealth/overviewcoph/faculty/xiaming-liu>

Featuring **Dr. Xiaoming Liu**
Department of Global Health
College of Public Health



Dr. Xiaoming Liu joined USF in 2018 as an Associate Professor in the Department of Global Health, College of Public Health. He received a BS and a MS in genetics from Fudan University, China, and a PhD in genetics with an emphasis on population genetics from The University of Texas Graduate School of Biomedical Sciences. His research interests lie in understanding the variants observed in human genomes. He develops novel statistical and computational methods to analyze DNA sequence data from three perspectives: population genetics, bioinformatics and genetic epidemiology. In population genetics, he developed the stairway plot method, a model-flexible method to estimate population demographic history using neutral SNP frequency spectra (SFS). His current research focus is inferring prehistoric demographic events of human populations using SFS and IBD segments. In bioinformatics, he developed a series of tools to facilitate functional annotation and pathogenetic prediction of human DNA variants, including dbNSFP, a functional annotation database for non-synonymous SNVs, and WGSa, a whole genome functional annotation pipeline. In genetic epidemiology, his research interest is utilizing functional annotation and pathogenetic prediction to improve the power of genotype-phenotype association analysis. His research interests also large-scale sequencing based Mendelian disease study, quantitative genetics of human traits and Darwinian medicine.