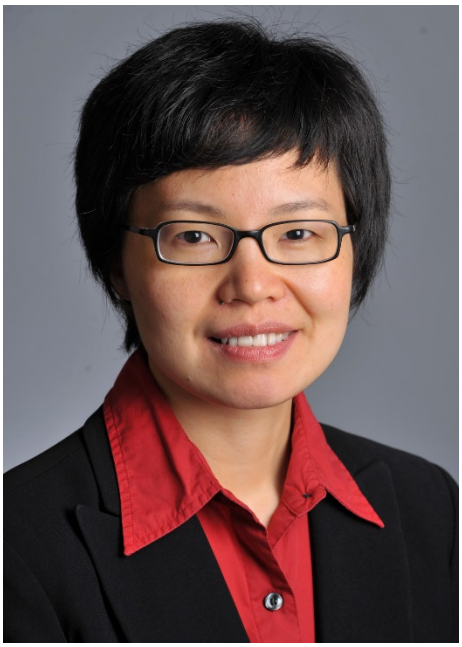
Institute for Biomedical Informatics

Seminar Series

Monday, March 26, 2018
12:00-1:00
014 CTW (Charles T. Wethington Building)



Deep phenotyping on EHR narratives facilitates genetic diagnosis by clinical exomes

Chunhua Weng, PhD
Columbia University

Abstract

Integration of detailed phenotypic information with genetic data is well established to facilitate accurate diagnosis of hereditary disorders. As a rich source of phenotypic information, electronic health records (EHRs) have the potential to empower diagnostic variant interpretation. However, how to accurately and efficiently extract phenotypes from heterogeneous EHR narratives remains a challenge. In this talk, I will describe a high-throughput EHR phenotype extraction and analysis framework that performs Human Phenotype Ontology (HPO) concept extraction and normalization from EHR narratives and prioritizes disease genes based on the HPO-coded phenotypic manifestations. Our results on four retrospective cohorts from multiple institutions show the promise of leveraging EHR data to automate phenotype-driven analysis of clinical exomes or genomes, facilitating the implementation of genomic medicine on scale.

Short Bio

Dr. Chunhua Weng is a tenured Associate Professor of Biomedical Informatics at Columbia University and an elected fellow of the American College of Medical Informatics (ACMI). She also co-leads the Biomedical Informatics Resource for the Columbia CTSA (The Irving Institute for Clinical and Translational Science). Dr. Weng holds a Ph.D. in Biomedical and Health Informatics from University of Washington at Seattle. Dr. Weng has been an active researcher in the field of Clinical Research Informatics since 2000 and has published extensively on natural language processing and knowledge representation for clinical research eligibility criteria, EHR data quality assessment and data analytics, and high-throughput EHR phenotyping.