UTHealth
The University of Texas Health Science Center at Houston
School of Biomedical Informatics

CPH Seminar in Precision Medicine

Annotating Regulatory Variants for Precision Medicine

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In the past ten years, Genome-wide association studies (GWAS), whole genome sequencing (WGS) and whole exome sequencing (WES) have identified tens of thousands of genetic variants that are associated with human diseases. Surprisingly, the majority (>90%) of these variants are located in regions that do not code for a protein, and currently annotated as Variants of Unknown Significances (VUS). Many of them are in the regions that harbor regulatory elements (such as promoters and enhancers), which affect the target gene expression through interaction with transcription factors (TFs). Mayo Clinic has established Center for Individualized Medicine (CIM) in 2008 to bring genomic discoveries to patient care, and was funded for the first Precision Medicine Initiative (PMI) project by NIH. I will introduce CIM initiatives in metagenomics, bioinformatics and pharmacogenomics and how they impact patients. I will discuss two methods we recently developed in detecting and prioritizing regulatory variants: 1) an ensemble method for GV function annotation. The method incorporates eight different tools, including CADD, GWAVA, Funseq, GWAS3D, SuRFR, DANN and fathmm-MKL) with a Bayes factor composite model. 2) A model that combines several epigenetic/chromatin features to improve regulatory variants' function prediction in tissue/cell type specific manner. Both methods are publicly available.

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