MIT Bioinformatics Seminar

Scalable Integrative Analysis of Large Biobank and Population-Based Whole Genome Sequencing Studies With Variant Functional Annotations

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Whole Genome/Exome Sequencing (WGS/WES) data and Electronic Health Records (EHRs), such as large scale national and institutional biobanks, have emerged rapidly worldwide. In this talk, I will discuss the analytic tools and resources for scalable analysis of large scale biobank- and population-based Whole Genome Sequencing (WGS) association studies of common and rare variants by integrating WGS data with multi-faceted functional annotation data. I will also provide a demo of FAVOR (favor.genohub.org), a variant functional annotation online portal and resource that provides multi-faceted functional annotations of genome-wide 9 billion variants, and FAVORAnnotator, a tool to functionally annotate any WGS/WES studies. Cloud-based platforms for these resources will be discussed. The presentation will be illustrated using ongoing large scale population-based whole genome sequencing studies and biobanks of quantitative, case-control, and time-to-event phenotypes, including

the Genome Sequencing Program (GSP) of the National Human Genome Research Institute and the Trans-Omics Precision Medicine Program (TOPMed) from the National Heart, Lung and Blood Institute, and the UK Biobank and FinnGen, which have collectively sequenced about 1 million genomes.