47. Using Genetics and Omics data to Inform Human Diseases

1 unit, Kushal Dey, November 5, 2025

Genome-wide association studies have identified >400,000 variants as potential risk factors for complex diseases and traits. Nearly 2/3rd of the approved drug targets in 2021 were supported by genetic studies. In this talk, we will cover statistical genetic approaches to analyzing GWAS data.

First, we will discuss the concepts and strategies to assess the genetic basis of disease risk by accounting for linkage disequilibrium and population stratification – including disease heritability and fine-mapping analysis to identify causal variants. Second, we will discuss how functional omics data – encompassing single-cell RNA-seq, ATAC-seq, Perturb-seq, and Spatially Resolved Transcriptomics (SRT) can be integrated with GWAS data to identify target genes of action for GWAS variants as well as pathways of active disease interest and informing drug intervention experiments. Third, we will discuss how omics-based quantitative trait loci (QTL) mapping studies can be integrated seamlessly with disease GWAS studies to inform variant-level assessment of function. Finally, we will discuss some of the ongoing and future directions that may be of scientific interest in this space.

Discussion Paper: Song, L., Chen, W., Hou, J., Guo, M. and Yang, J., 2025. Spatially resolved mapping of cells associated with human complex traits. Nature, 641(8064), pp.932-941. PMID: 40108460

Review Paper: IGVF Consortium. Deciphering the impact of genomic variation on function. Nature. 2024 Sep;633(8028):47-57. doi: 10.1038/s41586-024-07510-0. Epub 2024 Sep 4. PMID: 39232149