

Statistical Learning in the Genomic Era: Prediction, Causality, and Complex Diseases

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Abstract

Recent advances in human genetics and the rapid growth of large-scale biomedical data have created new opportunities for understanding the genetic architecture of complex diseases and advancing precision medicine. Genome-wide association studies (GWAS) have identified thousands of genetic variants associated with complex traits and diseases, providing important insights into their underlying biological mechanisms. However, translating these genetic associations into clinically meaningful prediction and causal understanding remains a major statistical challenge.

Polygenic risk scores (PRS) aggregate the effects of multiple genetic variants across the genome to quantify individual genetic susceptibility and have emerged as a promising tool for disease risk stratification, early screening, and personalized prevention. Beyond prediction, Mendelian randomization (MR) provides a statistical framework for investigating potential causal relationships between modifiable exposures and disease outcomes, thereby helping bridge the gap between genetic association and causal inference.

At the same time, machine learning methods offer powerful approaches for analyzing high-dimensional biomedical data. Techniques such as LASSO, random forests, and gradient boosting can facilitate feature selection, capture complex patterns, and integrate information from genetic variants, biomarkers, and clinical characteristics. When combined with conventional statistical genetic approaches, these methods can improve risk prediction and provide complementary insights into disease mechanisms.

In this talk, I will introduce an integrated framework that connects **genetic association, disease prediction, and causal inference** through GWAS, PRS, MR, and machine learning. Using large-scale genomic and biomedical data as examples, I will discuss how statistical learning can transform complex, high-dimensional data into interpretable evidence for disease risk assessment and biological discovery. Together, these approaches illustrate the expanding role of statistics in the genomic era and their potential to support more effective strategies for disease prevention and precision medicine.