

Indian Statistical Institute

Applied Statistics Unit

SEMINAR NOTICE

Speaker: Kushal De, Harvard School of Public Health

Title: Identifying disease-critical variants, genes and cell types using genetic and genomic data

Date: 23 August, 2022

Time: 16:15 PM

Venue: ASU Seminar Room

Online Platform: Google Meet (meet.google.com/nbb-kdit-dkr)

Abstract:

We are seeing an explosion in the size and scope of functional genomic and epigenomic data, raising the prospects for understanding the functional regulatory architecture underlying disease loci implicated by GWAS. Here, I present computational approaches that integrate large collections of functional data with disease GWAS to infer which variants, genes and cell types that are critical for disease. First, I will demonstrate how base-pair resolution functional data from deep learning models can be used to functionally resolve GWAS loci. Second, I will propose a strategy (*sc-linker*) to integrate gene programs characterizing enhancer-driven regulation and cell-type specificity of expression from single-cell RNA-seq data, enhancer-gene maps and GWAS summary statistics to infer underlying cell types and processes by which genetic variants affect disease. This approach captures the specificity in enrichment of blood cell types for blood biomarker traits (Erythroid for RBC count), and highlights novel associations, including GABA neurons for Major depressive disorder and disease microglia for Alzheimers' disease. Third, I will discuss a causal inference-based model that uses disease case-control data to identify genes causally altered in expression in disease state versus healthy and how much heritability signal is encapsulated in these genes.

All are invited to attend.