

**Speaker:** Po-Ru Loh

**Affiliations:** Brigham and Women's Hospital and Harvard Medical School

**Title:**

Uncovering hidden genetic variants and their effects on human traits

**Abstract:**

Genetic association studies over the past 15 years have discovered hundreds of thousands of common single-nucleotide polymorphisms (SNPs) associated with human phenotypes. However, common SNPs constitute only one type of genetic variation -- the easiest class to explore using existing analysis pipelines. In this talk, I will describe analyses of several other classes of genetic variation -- mosaic copy-number variants (CNVs), rare coding variants, and variable number tandem repeats (VNTRs) -- which have revealed many variants with large, readily-interpretable effects on human traits. These variants have traditionally been difficult to study in large cohorts, in which such variants have not been directly assayed. To overcome this challenge, we have developed statistical algorithms that leverage widely available SNP-array and exome-sequencing data by modeling haplotype-sharing among distantly related individuals.